

QUESTION

Which of the following is an example of facilitated diffusion across a cell membrane?

A Glucose transport via GLUT4

CORRECT

B Sodium-potassium pump

C Endocytosis

D Water movement through aquaporins

RIGHT ANSWER

OK A. Glucose transport via GLUT4

WHY THIS IS RIGHT

Facilitated diffusion is a type of passive transport in which molecules move down their concentration gradient across the cell membrane with the help of specific carrier or channel proteins, without the use of ATP.

Glucose transport via GLUT4 is a classic example of facilitated diffusion. GLUT4 is an insulin-regulated glucose transporter found mainly in skeletal muscle and adipose tissue. After insulin stimulation, GLUT4 transporters move to the cell membrane and allow glucose to enter the cell along its concentration gradient without energy expenditure.

Why other options are incorrect:

- Sodium-potassium pump: This is primary active transport that uses ATP to move Na⁺ out and K⁺ into the cell against their gradients.
- Endocytosis: A vesicular transport mechanism that requires energy and is not diffusion.
- Water movement through aquaporins: This is osmosis through channel proteins, not carrier-mediated facilitated diffusion.

QUESTION

Mechanism of action of botulinum toxin is:

- A** Inhibits NM receptor
- B** Inhibits reuptake of acetyl choline
- C** **Inhibits release of acetylcholine from lower motor neurons**
- D** Inhibits synthesis of acetyl choline

CORRECT

RIGHT ANSWER

- OK** **C. Inhibits release of acetylcholine from lower motor neurons**

WHY THIS IS RIGHT

Botulinum toxin blocks neuromuscular transmission by inhibiting the release of acetylcholine (ACh) at the presynaptic terminals of lower motor neurons.

Mechanism:

- Botulinum toxin cleaves SNARE proteins (e.g., synaptobrevin, SNAP-25).
- These proteins are essential for fusion of synaptic vesicles with the presynaptic membrane.
- Their inhibition prevents exocytosis of acetylcholine, leading to failure of neuromuscular transmission.

Clinical correlation:

- Causes descending flaccid paralysis (e.g., botulism)
- Used therapeutically in conditions like dystonia, spasticity, and cosmetic procedures

Why other options are incorrect:

- Inhibits NM receptor: Seen with drugs like curare, not botulinum toxin.
- Inhibits reuptake of acetylcholine: ACh is not reuptaken like monoamines; it is degraded by acetylcholinesterase.
- Inhibits synthesis of acetylcholine: Not the mechanism of botulinum toxin.

QUESTION

Which of the following is seen in low insulin: glucagon ratio?

- A** Activation of lipoprotein lipase
- B** Activation of glycogen synthase
- C** Activation of phosphofructokinase 1

D Activation of hormone-sensitive lipase

CORRECT

RIGHT ANSWER

OK D. Activation of hormone-sensitive lipase

WHY THIS IS RIGHT

A low insulin: glucagon ratio represents the fasting/starvation state, where the body shifts from energy storage to energy mobilization.

Metabolic effects of low insulin-high glucagon state:

- ↑ Lipolysis in adipose tissue
- Activation of hormone-sensitive lipase (HSL) → breakdown of triglycerides into free fatty acids and glycerol
- Fatty acids are used for β-oxidation and ketone body formation

Why other options are incorrect:

- Lipoprotein lipase: Activated by insulin (fed state) for fat storage
- Glycogen synthase: Active in fed state (high insulin) for glycogen formation
- Phosphofructokinase-1 (PFK-1): Activated in glycolysis (fed state), not during fasting

QUESTION

A diabetic patient was advised exercise by the treating physician. The glucose transporter which is insulin sensitive is:

A Glut 1

B Glut 2

C Glut 3

D Glut 4

CORRECT

RIGHT ANSWER

OK D. Glut 4

WHY THIS IS RIGHT

GLUT-4 is the insulin-sensitive glucose transporter, primarily found in:

- Skeletal muscle
- Adipose tissue

Mechanism:

- In response to insulin, GLUT-4 translocates from intracellular vesicles to the cell membrane
- This increases glucose uptake into cells
- Exercise can also stimulate GLUT-4 translocation independent of insulin

Clinical relevance in diabetes:

- Exercise improves glucose uptake even when insulin action is impaired
- Hence, exercise is beneficial in diabetes management

Why other options are incorrect:

- GLUT-1: RBCs, brain (insulin-independent)
- GLUT-2: Liver, pancreas (high K_m , bidirectional)
- GLUT-3: Neurons (high affinity, insulin-independent)

QUESTION

A miner has been trapped in a tunnel for the past 5 days without access to food. What is the primary source of energy for the brain under these conditions?

A Glycogenolysis

B Ketosis

CORRECT

C Gluconeogenesis

D Lipolysis

RIGHT ANSWER

OK B. Ketosis

WHY THIS IS RIGHT

During prolonged fasting/starvation ($\geq 3-5$ days), the body adapts to conserve glucose. The brain, which normally depends on glucose, shifts to using ketone bodies as its primary energy source.

Time Period	Main Energy Source	Brain Fuel
0-18 hrs	Hepatic glycogenolysis	Glucose
18-48 hrs	Gluconeogenesis (AA, glycerol, lactate)	Glucose
>48 hrs / 5 days	Ketogenesis	Ketone bodies

QUESTION

Entry of glucose is insulin independent in all of them except:

A RBC

B Pancreas

C Adipose tissues

CORRECT

D Brain

RIGHT ANSWER

OK C. Adipose tissues

WHY THIS IS RIGHT

Major Glucose transporters and their function is summarized as:

Transporter	Location	Function
Glut-1	Brain, Kidney, Placenta, Erythrocytes, Cancer	Basal uptake of glucose, High affinity
Glut-2	Liver, Pancreatic cell, small intestine	Fed state
Glut-3	Brain, Kidney, Placenta	Glucose uptake, Maximum affinity for glucose
Glut-4	Heart, Skeletal muscle, Adipose tissue	Insulin-stimulated glucose uptake
Glut-5	Small intestine, Sperms	Fructose

QUESTION

| A patient diagnosed with diabetes has high increased LDL and VLDL because of ?

- A** Hormone sensitive lipase and lipoprotein lipase both increased
- B** **Hormone sensitive lipase increased & lipoprotein lipase decreased**
- C** Lipoprotein lipase increased & hormone sensitive lipase decreased
- D** Lipoprotein lipase and hormone sensitive lipase both decreased

CORRECT

RIGHT ANSWER

- OK** **B. Hormone sensitive lipase increased & lipoprotein lipase decreased**

WHY THIS IS RIGHT

In diabetes mellitus (especially uncontrolled diabetes), insulin deficiency or resistance alters lipid metabolism, leading to hypertriglyceridemia and increased VLDL and LDL levels.

1\ Effect on Hormone-Sensitive Lipase (HSL)

- Insulin normally inhibits HSL in adipose tissue.
- When insulin is deficient -> HSL activity increases.
- This causes enhanced lipolysis in adipose tissue, releasing large amounts of free fatty acids (FFA) into the circulation.
- The liver takes up these FFAs and converts them into triglycerides, which are packaged as VLDL -> increased VLDL production.

2\ Effect on Lipoprotein Lipase (LPL)

- Insulin normally stimulates LPL, which hydrolyzes triglycerides in VLDL and chylomicrons to allow uptake into tissues.
- In diabetes -> LPL activity decreases due to lack of insulin.
- This results in reduced clearance of VLDL and chylomicrons, further increasing plasma triglycerides.

QUESTION

Which of the following tissues is unable to take up glucose in the absence of insulin?

A Skeletal muscles

CORRECT

B RBC

C Brain

D Kidney

RIGHT ANSWER

OK A. Skeletal muscles

WHY THIS IS RIGHT

Glucose uptake in different tissues depends on specific glucose transporters (GLUTs), some of which are insulin dependent.

- Skeletal muscle and adipose tissue require insulin for glucose entry.
- Insulin stimulates translocation of GLUT-4 transporters to the cell membrane.
- Without insulin, GLUT-4 does not move to the membrane, so glucose uptake is markedly reduced.

Therefore, skeletal muscle cannot effectively take up glucose in the absence of insulin.

Some tissues use transporters that do not require insulin:

- RBCs -> GLUT-1
- Brain -> GLUT-1 and GLUT-3
- Kidney, liver, pancreatic β -cells -> GLUT-2

These tissues can take up glucose even during fasting or insulin deficiency.

QUESTION

In a 90 kg man following a meal, which of the following enzymes or intermediates is most likely activated to promote anabolic processes contributing to weight gain?

1. Malonyl CoA
2. Acetyl CoA carboxylase
3. Mitochondrial carnitine transporter
4. Tricarboxylate transporter (citrate)

A 1, 2 and 4

CORRECT

B 1 and 2

C 1 and 3

D 1, 2, 3 and 4

RIGHT ANSWER

OK A. 1, 2 and 4

WHY THIS IS RIGHT

In the fed state (post-meal), insulin promotes anabolic metabolism, especially fatty acid synthesis and storage, contributing to weight gain.

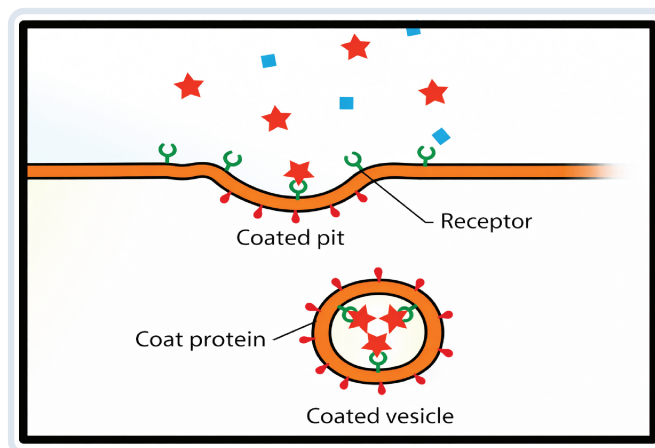
Key Activated Components:

- Acetyl-CoA carboxylase (ACC): Activated by insulin → converts acetyl-CoA to malonyl-CoA (rate-limiting step of fatty acid synthesis).
- Malonyl-CoA: Promotes lipogenesis and inhibits fatty acid oxidation.
- Tricarboxylate transporter (citrate shuttle): Moves citrate from mitochondria to cytosol to provide acetyl-CoA for fatty acid synthesis.

Malonyl-CoA inhibits the mitochondrial carnitine transporter (CPT-1), preventing fatty acid entry into mitochondria for β -oxidation.

QUESTION

Based on the image provided, which process is being depicted?



- A Transcytosis
- B Clathrin mediated endocytosis**
- C Exocytosis
- D Pinocytosis

CORRECT

RIGHT ANSWER

- B. Clathrin mediated endocytosis**

WHY THIS IS RIGHT

The image depicts vesicle formation at the plasma membrane coated with a protein lattice (clathrin), which selectively internalizes receptor-bound molecules into the cell. Clathrin-mediated endocytosis is a highly specific form of receptor-mediated uptake of ligands such as LDL, transferrin, and hormones.

Why the others are incorrect:

- A. Transcytosis: Involves vesicular transport across a cell, from one side to the other, not selective vesicle budding at the membrane.
- C. Exocytosis: Refers to vesicles fusing with the plasma membrane to release contents, opposite of endocytosis.
- D. Pinocytosis: Non-specific uptake of extracellular fluid and solutes without clathrin-coated vesicles.

QUESTION

During periods of prolonged fasting, one of the following tissues preferentially uses fatty acids as energy-providing substrates. It is because this tissue has the highest concentration of lipoprotein lipase. Identify this tissue:

A Adipose tissue

B Skeletal muscle

C Liver

D Cardiac muscle

CORRECT

RIGHT ANSWER

OK D. Cardiac muscle

WHY THIS IS RIGHT

During prolonged fasting, cardiac muscle preferentially uses fatty acids for energy because it has high concentrations of lipoprotein lipase (LPL), which hydrolyzes circulating triglycerides into free fatty acids for uptake. Fatty acids are an efficient fuel for the heart, providing sustained energy without relying on glucose.

Why the others are incorrect:

- A. Adipose tissue: Mainly stores fat; it releases fatty acids during fasting but does not use them preferentially for energy.
- B. Skeletal muscle: Uses both glucose and fatty acids, but cardiac muscle oxidizes fatty acids more efficiently.
- C. Liver: Primarily performs gluconeogenesis and ketone body production; it does not rely on fatty acid oxidation for its own energy.

QUESTION

A 14-year-old boy is brought to the emergency department due to excessive urination and thirst. He has lost 4.5 kg in the last 3 weeks. Physical examination shows dry mucous membranes. Laboratory studies reveal blood glucose of 455 mg/dl, normal anion gap, and hemoglobin A1c of 11.3%. The patient is diagnosed with type 1 diabetes, and treatment with insulin is initiated. In addition to lowering blood glucose, insulin increases glycogen synthesis in hepatocytes. Activation of which of the following molecules most likely promotes this metabolic effect?

A Janus kinase (JAK)

B Phospholipase C

C Protein kinase A

D Protein phosphatase

CORRECT

RIGHT ANSWER

OK

D. Protein phosphatase

WHY THIS IS RIGHT

Insulin promotes glycogen synthesis in hepatocytes by activating protein phosphatases, particularly protein phosphatase 1 (PP1). PP1 dephosphorylates and activates glycogen synthase while inactivating glycogen phosphorylase, shifting metabolism toward glycogen storage. This effect complements insulin's role in lowering blood glucose.

Why the others are incorrect:

- A. Janus kinase (JAK): Mediates cytokine and growth hormone signaling, not insulin's glycogen effects.
- B. Phospholipase C: Part of Gq-coupled receptor pathways, involved in IP3/DAG signaling, not glycogen synthesis.
- C. Protein kinase A: Activated by cAMP in response to glucagon/epinephrine; phosphorylates glycogen synthase to inhibit glycogen formation, opposite of insulin's action.

QUESTION

Match the cell organelle with its marker enzymes:

1. Plasma membrane	a. Galactosyltransferase
2. Golgi bodies	b. Lactate dehydrogenase
3. Cytosol	c. Acid phosphatase
4. Lysosomes	d. 5'-nucleotidase

- A** 1-a, 2-b, 3-c, 4-d
- B** 1-a, 2-c, 3-b, 4-d
- C** 1-d, 2-a, 3-b, 4-c CORRECT
- D** 1-d, 2-c, 3-a, 4-b

RIGHT ANSWER

OK C. 1-d, 2-a, 3-b, 4-c

WHY THIS IS RIGHT

- Plasma membrane (1 -> D): 5'-nucleotidase is a marker enzyme for the plasma membrane, involved in nucleotide metabolism.
- Golgi bodies (2 -> A): Galactosyltransferase is localized in the Golgi apparatus and is used as a marker for its presence.
- Cytosol (3 -> B): Lactate dehydrogenase (LDH) is abundant in the cytosol, catalyzing conversion of pyruvate ↔ lactate.
- Lysosomes (4 -> C): Acid phosphatase is a lysosomal enzyme, used as a marker for lysosomal content.

Why other options are incorrect:

- Mixing these enzymes with the wrong organelles would misidentify cellular localization, e.g., LDH is not in lysosomes, and acid phosphatase is not cytosolic.
- Each marker enzyme is specific to its organelle for subcellular fractionation studies.

QUESTION

| Choose the right sequence of steps in the conversion of cholesterol to testosterone:

A 20, 22 desmolase, 3 β -hydroxysteroid dehydrogenase 17 alpha hydroxylase, 17, 20 lyase

CORRECT

B 3 β -hydroxysteroid dehydrogenase 17 alpha hydroxylase, 17, 20 lyase, 20, 22 desmolase

C 20, 22 desmolase, 17, 20 lyase, 17 alpha hydroxylase, 3 β -hydroxysteroid dehydrogenase

D 17 alpha hydroxylase, 3 β -hydroxysteroid dehydrogenase, 20, 22 desmolase

RIGHT ANSWER

OK **A. 20, 22 desmolase, 3 β -hydroxysteroid dehydrogenase 17 alpha hydroxylase, 17, 20 lyase**

WHY THIS IS RIGHT

The source quiz contains the placeholder "\$19" here and does not include a written explanation for this item.

QUESTION

| 2,3 BPG causes unloading of oxygen to tissues because:

- A** It decreases ATP production and hence breaks the links between globin chains
- B** It increases ATP production
- C** **Binds to the central cavity of hemoglobin surrounded by histidine and lysine and favours salt bridge formation** CORRECT
- D** Inhibits Taut structure formation

RIGHT ANSWER

- OK** **C. Binds to the central cavity of hemoglobin surrounded by histidine and lysine and favours salt bridge formation**

WHY THIS IS RIGHT

2,3-Bisphosphoglycerate (2,3-BPG) binds to the central cavity of deoxyhemoglobin, interacting with positively charged residues (histidine and lysine). This stabilizes the T (tense) state of hemoglobin, which has lower affinity for oxygen, thereby promoting oxygen unloading to tissues.

Why other options are incorrect:

- A. Decreases ATP production: Not related to hemoglobin-oxygen affinity.
- B. Increases ATP production: Irrelevant; 2,3-BPG modulates oxygen binding, not energy production.
- D. Inhibits Taut structure formation: False; 2,3-BPG stabilizes the T (tense) form, favoring oxygen release.

QUESTION

A TPN bag consists of 100g glucose, 30g amino acids, and 40g lipids. What is the amount of calories delivered?

A 840 Kcal

B 880 Kcal

CORRECT

C 640 Kcal

D 680 Kcal

RIGHT ANSWER

B. 880 Kcal

WHY THIS IS RIGHT

- In patients who cannot (unconscious; removal of large part of gut) or should not (major trauma or surgery) use their gastrointestinal tract, total parenteral feeding has to be resorted.
- The solution may be infused through one of the large vessels like subclavian vein or superior vena cava, where the blood flow is sufficient to dilute the hypertonic solution.
- This procedure is known as total parenteral nutrition (TPN) or intravenous hyperalimentation.
- The number of calories delivered by the given TPN (Total Parenteral Nutrition) bag is 880 Kcal.

The caloric values of different food components are:

- Carbohydrate (glucose) - 4KCal/g
- Proteins (amino acids) - 4.2KCal/g = ~4KCal/g
- Fat (lipids) - 9Kcal/g

Calculation of the number of calories:

- 100 gm of glucose = $100 \times 4 \text{ KCal} = 400 \text{ KCal}$
- 30 gm of amino acids = $30 \times 4 \text{ KCal} = 120 \text{ KCal}$
- 40 gm of lipids = $40 \times 9 \text{ KCal} = 360 \text{ KCal}$

Total amount of calories delivered = $400 + 120 + 360 = 880 \text{ Kcal}$

QUESTION

Which of the following enzymes will show reduced activity in a person who is fasting?

A Fructose 2,6 bisphosphatase

B Fructose 1,6 bisphosphatase

C Pyruvate carboxylase

D Phosphofructokinase-2

CORRECT

RIGHT ANSWER

OK D. Phosphofructokinase-2

WHY THIS IS RIGHT

During fasting, glucagon signaling leads to activation of cAMP-dependent protein kinase, which phosphorylates PFK-2, converting it into a fructose-2,6-bisphosphatase-active form. This reduces PFK-2 activity, lowering fructose-2,6-bisphosphate levels. Since fructose-2,6-bisphosphate is an allosteric activator of PFK-1, glycolysis is suppressed and gluconeogenesis is favored.

Why other options are incorrect:

- A. Fructose-2,6-bisphosphatase: Activity actually increases during fasting to promote gluconeogenesis.
- B. Fructose-1,6-bisphosphatase: Activity increases during fasting to drive gluconeogenesis.
- C. Pyruvate carboxylase: Activity increases to supply oxaloacetate for gluconeogenesis.

QUESTION

Match the following transporters with the molecule they carry:

1. Hemopexin	a. Thyroxin
2. Haptoglobin	b. Free fatty acid
3. Pre-albumin	c. Heme
4. Albumin	d. Haemoglobin

A 1b, 2a, 3c, 4d

B 1c, 2d, 3a, 4b

CORRECT

C 1a, 2c, 3b, 4d

D 1c, 2a, 3d, 4b

RIGHT ANSWER

OK B. 1c, 2d, 3a, 4b

WHY THIS IS RIGHT

- Hemopexin (1 → C): Binds free heme in plasma to prevent oxidative damage and deliver it to the liver for degradation.
- Haptoglobin (2 → D): Binds free hemoglobin released from erythrocytes, facilitating clearance by the reticuloendothelial system.
- Prealbumin (transthyretin) (3 → A): Transports thyroxine (T₄) and also carries retinol-binding protein.
- Albumin (4 → B): Major plasma protein that binds and transports free fatty acids, drugs, and other hydrophobic molecules.

Why other options are incorrect:

- Switching hemopexin and haptoglobin would mismatch heme vs hemoglobin binding.
- Prealbumin does not bind fatty acids-that is albumin's role.
- Albumin does not transport thyroxine efficiently; prealbumin/transthyretin does.

QUESTION

Identify the incorrect pair of UV absorption wavelength with respective compound:

- A** Tryptophan- 280 nm
- B** Nucleic acids - 260 nm (N-bases)
- C** NADH/NADPH - 340 nm
- D** Porphyrins - 200 nm

CORRECT

RIGHT ANSWER

- OK** **D. Porphyrins - 200 nm**

WHY THIS IS RIGHT

Porphyrins have a strong Soret band around 400-420 nm, not 200 nm. This allows their detection and quantification in biochemical assays.

Why other options are correct:

- A. Tryptophan - 280 nm: True; aromatic amino acids absorb strongly around 280 nm.
- B. Nucleic acids - 260 nm: True; nucleobases absorb UV light maximally at 260 nm.
- C. NADH/NADPH - 340 nm: True; reduced forms of nicotinamide coenzymes absorb at 340 nm.

QUESTION

| All of the following reactions may be inhibited in an alcoholic except:

A Glycolysis

B TCA

C Lactic acid production

CORRECT

D Fatty acid oxidation

RIGHT ANSWER

OK C. Lactic acid production

WHY THIS IS RIGHT

In chronic alcohol consumption, ethanol is metabolized to acetaldehyde and then acetate, producing excess NADH in the liver. This high NADH/NAD⁺ ratio inhibits:

- Glycolysis: Glyceraldehyde-3-phosphate dehydrogenase is inhibited due to NAD⁺ depletion.
- TCA cycle: High NADH inhibits key dehydrogenases (isocitrate, α -ketoglutarate, malate).
- Fatty acid oxidation: Requires NAD⁺; high NADH inhibits β -oxidation, promoting fatty liver.

Lactic acid production, however, is increased rather than inhibited, because excess NADH drives pyruvate $\rightarrow$ lactate via lactate dehydrogenase, often causing lactic acidosis.

Why other options are incorrect:

Glycolysis, TCA, and fatty acid oxidation are all directly inhibited by the altered NADH/NAD⁺ ratio in alcohol metabolism.

QUESTION

A 5-year-old girl was washing her doll with shampoo containing rotenone. Her mother noticed her in an unconscious state. Which is inhibited by the above chemical?

A NADH dehydrogenase

CORRECT

B Succinate dehydrogenase

C Cytochrome C

D Cytochrome oxidase

RIGHT ANSWER

OK **A. NADH dehydrogenase**

WHY THIS IS RIGHT

Rotenone is a pesticide that acts as a potent inhibitor of Complex I (NADH:ubiquinone oxidoreductase) in the mitochondrial electron transport chain. By blocking electron transfer from NADH to coenzyme Q, it prevents oxidative phosphorylation, leading to cellular ATP depletion and acute toxicity, which explains the child's unconscious state.

Why other options are incorrect:

- B. Succinate dehydrogenase: Complex II; not inhibited by rotenone.
- C. Cytochrome C: Electron carrier; rotenone acts upstream, not directly on cytochrome C.
- D. Cytochrome oxidase: Complex IV; inhibited by cyanide, not rotenone.

QUESTION

| All of the following are NADPH generating reactions except:

- A** 6-phospho gluconate dehydrogenase
- B** Cytoplasmic isocitrate dehydrogenase
- C** Malic enzyme
- D** **Ketoacyl-ACP dehydrogenase**

CORRECT

RIGHT ANSWER

- OK** **D. Ketoacyl-ACP dehydrogenase**

WHY THIS IS RIGHT

Ketoacyl-ACP dehydrogenase is part of fatty acid synthesis, catalyzing the reduction of β -ketoacyl-ACP to β -hydroxyacyl-ACP using NADPH, but it consumes NADPH rather than generating it.

Why other options are incorrect:

- A. 6-phosphogluconate dehydrogenase: Generates NADPH in the pentose phosphate pathway.
- B. Cytoplasmic isocitrate dehydrogenase: Produces NADPH by converting isocitrate $\rightarrow$ α -ketoglutarate in the cytosol.
- C. Malic enzyme: Converts malate $\rightarrow$ pyruvate, generating NADPH for fatty acid synthesis.

QUESTION

In an electron Transport system, which part will be the last to get electrons?

A Coenzyme Q

B FADH₂

C O₂

CORRECT

D Cytochrome C

RIGHT ANSWER

OK C. O₂

WHY THIS IS RIGHT

In the electron transport chain (ETC), electrons flow from NADH and FADH₂ → Complex I/II → Coenzyme Q → Complex III → Cytochrome C → Complex IV and finally to molecular oxygen (O₂), which acts as the terminal electron acceptor. Oxygen is reduced to water, completing the chain.

Why other options are incorrect:

- A. Coenzyme Q: Accepts electrons early from Complex I and II, not last.
- B. FADH₂: Donates electrons to Complex II, entering the chain before CoQ.
- D. Cytochrome C: Transfers electrons from Complex III to Complex IV; still before O₂.

QUESTION

Which of the following enzyme activity decreases in fasting?

A Hormone-sensitive lipase

B Glycogen phosphorylase

C Pyruvate carboxylase

D **Phosphofructokinase 1**

CORRECT

RIGHT ANSWER

OK **D. Phosphofructokinase 1**

WHY THIS IS RIGHT

During fasting, the body shifts toward glucose conservation and production, suppressing glycolysis. Phosphofructokinase-1 (PFK-1) is a key rate-limiting enzyme of glycolysis and is inhibited by low insulin, high glucagon, increased ATP, and citrate levels seen in fasting. Thus, its activity decreases to reduce glucose utilization.

Why other options are incorrect:

- A. Hormone-sensitive lipase: Activity increases in fasting to mobilize fatty acids from adipose tissue.
- B. Glycogen phosphorylase: Increases to promote glycogen breakdown for glucose supply.
- C. Pyruvate carboxylase: Increases to drive gluconeogenesis during fasting.

QUESTION

The enzyme that is common for both glycolysis and gluconeogenesis is?

- A** Pyruvate kinase
- B** Phosphofructokinase
- C** Phosphoenolpyruvate carboxykinase

D Phosphoglycerate kinase

CORRECT

RIGHT ANSWER

OK D. Phosphoglycerate kinase

WHY THIS IS RIGHT

Phosphoglycerate kinase catalyzes a reversible reaction converting 1,3-bisphosphoglycerate to 3-phosphoglycerate with ATP generation in glycolysis, and the reverse reaction in gluconeogenesis using ATP. Because the reaction is near equilibrium, the same enzyme functions in both pathways, unlike the irreversible regulatory steps that require bypass enzymes.

Why other options are incorrect:

- A. Pyruvate kinase: Irreversible glycolytic enzyme; bypassed by pyruvate carboxylase and PEPCK.
- B. Phosphofructokinase: Key irreversible glycolytic enzyme; bypassed by fructose-1,6-bisphosphatase.
- C. PEP carboxykinase: Used only in gluconeogenesis, not glycolysis.

QUESTION

| Which of the following best describes the amphibolic nature of the citric acid cycle?

A It generates ATP and produces citrate for fatty acid synthesis.

CORRECT

B It exclusively produces NADH for oxidative phosphorylation.

C It breaks down fatty acids and synthesises only cholesterol.

D It provides intermediates solely for glucose synthesis.

RIGHT ANSWER

OK **A. It generates ATP and produces citrate for fatty acid synthesis.**

WHY THIS IS RIGHT

The citric acid (TCA) cycle is amphibolic because it serves both catabolic and anabolic roles. Catabolically, it oxidizes acetyl-CoA to generate NADH, FADH₂, and GTP (ATP equivalent) for energy production. Anabolically, intermediates are withdrawn for biosynthesis-most notably citrate, which is exported to the cytosol and used for fatty acid and cholesterol synthesis.

Why other options are incorrect:

B. It exclusively produces NADH for oxidative phosphorylation: The TCA cycle does more than produce NADH; it also supplies anabolic intermediates.

C. It breaks down fatty acids and synthesises only cholesterol: Fatty acid breakdown occurs via β -oxidation, not the TCA cycle.

D. It provides intermediates solely for glucose synthesis: TCA intermediates contribute to multiple pathways, not only gluconeogenesis.

QUESTION

An infant came with vomiting, jaundice, and hypoglycemia. What is the most likely diagnosis?

A Galactosemia

CORRECT

B Phenylketonuria

C Wilson disease

D Menke disease

RIGHT ANSWER

A. Galactosemia

WHY THIS IS RIGHT

Galactosemia is an inborn error of metabolism caused by deficiency of galactose-1-phosphate uridylyltransferase, leading to accumulation of toxic metabolites after milk ingestion.

- Presents in neonates/infants after starting milk feeds
- Features include vomiting, jaundice, hypoglycemia, hepatomegaly, and failure to thrive
- Can also cause cataracts and risk of E. coli sepsis
- Early diagnosis is crucial
- Management requires immediate elimination of galactose/lactose from diet

Contrast:

- Phenylketonuria: intellectual disability, musty odor, no jaundice/hypoglycemia
- Wilson disease: presents later with liver and neurological features
- Menkes disease: copper metabolism disorder with kinky hair and neurodegeneration

QUESTION

A 1-week-old child was presented with early cataract. He also has refusal to feed, vomiting, lethargy, jaundice, and sepsis. This can be caused due to the deficiency of which enzyme?

A Catalase

B Galactose-1-phosphate uridylyltransferase

CORRECT

C Aldose reductase

D Hexokinase

RIGHT ANSWER

OK

B. Galactose-1-phosphate uridylyltransferase

WHY THIS IS RIGHT

Classic galactosemia is caused by deficiency of galactose-1-phosphate uridylyltransferase (GALT) and presents in neonates soon after milk feeding.

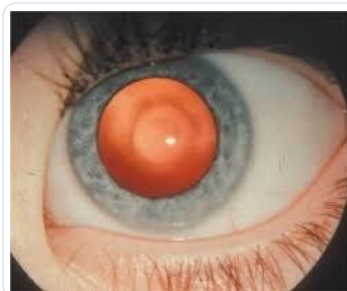
- Early cataract (due to galactitol accumulation in lens)
- Poor feeding, vomiting, lethargy
- Jaundice and hepatomegaly
- Failure to thrive
- Risk of E. coli sepsis

Biochemical basis:

- Block in conversion of galactose-1-phosphate → UDP-galactose
- Accumulation of galactose-1-phosphate (toxic) and galactitol

Why not others:

- Aldose reductase deficiency: Would reduce cataract formation (not cause disease)
- Hexokinase: General glycolysis enzyme, not specific
- Catalase: Involved in oxidative stress, unrelated



QUESTION

| Which of the following enzyme is deficient in Fabry's Disease?

A α -Galactosidase

CORRECT

B β -Galactosidase

C β -Glucosidase

D Sphingomyelinase

RIGHT ANSWER

OK **A. α -Galactosidase**

WHY THIS IS RIGHT

Fabry disease is a lysosomal storage disorder caused by deficiency of the enzyme α -galactosidase A. This enzyme normally breaks down globotriaosylceramide (ceramide trihexoside). When the enzyme is deficient, these glycosphingolipids accumulate in vascular endothelium, kidneys, heart, and nerves, leading to progressive organ damage.

Typical clinical features:

- Angiokeratomas (skin lesions)
- Acroparesthesia (burning pain in hands and feet)
- Hypohidrosis
- Progressive renal failure and cardiomyopathy

Why other options are incorrect:

- β -Galactosidase deficiency: Causes GM1 gangliosidosis.
- β -Glucosidase deficiency: Causes Gaucher disease.
- Sphingomyelinase deficiency: Causes Niemann-Pick disease.

QUESTION

A child presents with abdominal distension, fasting hypoglycemia, and hepatomegaly (liver palpable 8 cm below the costal margin). The liver is smooth on palpation. Which of the following enzyme deficiencies is the most likely cause?

A Glucose-6-phosphatase

CORRECT

B Glucose-6-phosphate dehydrogenase

C Myophosphorylase

D Phosphofructokinase

RIGHT ANSWER

OK

A. Glucose-6-phosphatase

WHY THIS IS RIGHT

Deficiency of glucose-6-phosphatase causes Glycogen Storage Disease Type I (Von Gierke disease). This enzyme normally converts glucose-6-phosphate to free glucose in the final step of glycogenolysis and gluconeogenesis in the liver and kidney.

When this enzyme is deficient:

- Glucose cannot be released into the bloodstream, leading to severe fasting hypoglycemia.
- Glucose-6-phosphate accumulates, which is diverted into glycogen synthesis, causing excess glycogen deposition in the liver.
- This results in marked hepatomegaly and abdominal distension.

Why other options are incorrect:

- Glucose-6-phosphate dehydrogenase deficiency: Causes hemolytic anemia due to impaired NADPH production in RBCs, not hepatomegaly with hypoglycemia.
- Myophosphorylase deficiency: Causes McArdle disease, presenting with exercise intolerance and muscle cramps, not liver enlargement.
- Phosphofructokinase deficiency: Causes Tarui disease (GSD VII) with muscle weakness and hemolysis.

QUESTION

A 2-week-old exclusively breastfed infant is brought with poor feeding, vomiting, lethargy, and failure to thrive. On examination, the infant has hepatomegaly and bilateral cataracts. Laboratory evaluation shows hypoglycemia and elevated liver enzymes. Which of the following enzyme deficiencies is most likely responsible for this presentation?

A Galactokinase

B Galactose-1-phosphate uridyl transferase (GALT)

CORRECT

C Aldolase B

D Fructokinase

RIGHT ANSWER

OK B. Galactose-1-phosphate uridyl transferase (GALT)

WHY THIS IS RIGHT

Classic galactosemia is caused by deficiency of galactose-1-phosphate uridyl transferase (GALT), an enzyme involved in the metabolism of galactose. This condition commonly presents in breastfed infants, because breast milk contains lactose, which is broken down into glucose and galactose.

When GALT is deficient, galactose-1-phosphate and galactose accumulate in tissues such as the liver, brain, and lens of the eye. Excess galactose is converted to galactitol, which accumulates in the lens and causes cataracts. Accumulation in the liver leads to hepatomegaly and liver dysfunction, while toxic metabolites contribute to lethargy and feeding difficulties.

Why other options are incorrect:

- Galactokinase deficiency: Causes cataracts only, without severe liver involvement or systemic toxicity.
- Aldolase B deficiency: Seen in hereditary fructose intolerance, presenting after fructose ingestion.
- Fructokinase deficiency: Causes essential fructosuria, a benign condition.

QUESTION

| Rice water diarrhoea, intoxication is due to which ganglioside?

A GM-1 ganglioside receptor

CORRECT

B GM-2 ganglioside receptor

C GM-3 ganglioside receptor

D GD-1a ganglioside receptor

RIGHT ANSWER

OK

A. GM-1 ganglioside receptor

WHY THIS IS RIGHT

Rice water diarrhea" is characteristic of cholera, caused by *Vibrio cholerae*. The cholera toxin specifically binds to the GM1 ganglioside receptor present on the intestinal epithelial cell membrane.

- Cholera toxin binds to GM1 ganglioside on enterocytes.
- The toxin activates adenylate cyclase via Gs protein stimulation.
- This leads to ↑ cAMP levels.
- Increased cAMP causes excess secretion of chloride and water into the intestinal lumen.

Result:

- > Profuse watery diarrhea ("rice water stools")
- > Severe dehydration and electrolyte imbalance

Why other options are incorrect:

- GM2 ganglioside: Associated with Tay-Sachs disease metabolism, not toxin binding.
- GM3 ganglioside: Not involved in cholera toxin action.
- GD1a ganglioside: Involved in other neuronal interactions, not cholera.

QUESTION

A Mediterranean person didn't receive primaquine. Which pathway is affected by G-6-PD enzyme deficiency?

A HMP pathway

CORRECT

B Glycolysis

C Gluconeogenesis

D Kreb cycle

RIGHT ANSWER

OK **A. HMP pathway**

WHY THIS IS RIGHT

Glucose-6-phosphate dehydrogenase (G6PD) is the key regulatory enzyme of the Hexose Monophosphate (HMP) pathway / Pentose Phosphate Pathway.

This pathway generates NADPH, which is essential for:

- Maintaining reduced glutathione (GSH) in red blood cells
- Protecting RBCs from oxidative damage

In G6PD deficiency (common in Mediterranean populations):

- ↓ NADPH → ↓ reduced glutathione
- RBCs become vulnerable to oxidative stress (e.g., drugs like primaquine, infections, fava beans)
- Leads to hemolysis

Why other options are incorrect:

- Glycolysis: Generates ATP, not primarily dependent on G6PD
- Gluconeogenesis: Produces glucose, unrelated to G6PD
- Krebs cycle: Occurs in mitochondria, not affected by G6PD deficiency

QUESTION

| True about Warburg effect?

A Makes cancer cells immortal

B Aerobic glycolysis with lactate formation

CORRECT

C Produce more ATP

D Decrease glucose uptake

RIGHT ANSWER

OK B. Aerobic glycolysis with lactate formation

WHY THIS IS RIGHT

The Warburg effect refers to the preference of cancer cells to utilize aerobic glycolysis, meaning they convert glucose to lactate even in the presence of adequate oxygen.

Normally: Oxygen present -> cells use oxidative phosphorylation (high ATP yield)

In cancer cells: Despite oxygen -> glycolysis predominates -> lactate production

Why does this happen?

- Provides rapid ATP (though less efficient)
- Generates intermediates for biosynthesis (nucleotides, lipids, amino acids) needed for rapid cell proliferation

Why other options are incorrect:

- Makes cancer cells immortal: Immortality is due to telomerase activation, not Warburg effect
- Produce more ATP: Glycolysis produces less ATP than oxidative phosphorylation
- Decrease glucose uptake: Cancer cells actually show increased glucose uptake (basis of PET scan)

QUESTION

Cherry red spots on retina and mental retardation is seen in:



A Gaucher's disease

B **Tay Sach's disease**

CORRECT

C Hunter syndrome

D Krabbe' disease

RIGHT ANSWER

OK **B. Tay Sach's disease**

WHY THIS IS RIGHT

A cherry red spot on the retina with neurodegeneration (mental retardation) is characteristic of Tay-Sachs disease, a lysosomal storage disorder caused by deficiency of hexosaminidase A.

- Deficiency of hexosaminidase A -> accumulation of GM2 ganglioside in neurons
- Leads to progressive neurodegeneration
- The surrounding retina becomes pale due to lipid accumulation, while the fovea appears bright red (cherry red spot) because it lacks ganglion cells

Important distinguishing feature:

- No hepatosplenomegaly (helps differentiate from Niemann-Pick disease, which also has cherry red spot but with organomegaly)

Why other options are incorrect:

- Gaucher disease: Hepatosplenomegaly, bone crises (no cherry red spot)
- Hunter syndrome: Mucopolysaccharidosis with coarse facies, no cherry red spot
- Krabbe disease: Demyelination with globoid cells, no cherry red spot

QUESTION

A 16-year-old boy presents with exercise intolerance, early fatigue, and painful muscle cramps during strenuous activity. He reports that symptoms improve with rest. There is no significant rise in serum lactate levels after exercise testing. Serum creatine kinase is mildly elevated. Which of the following enzyme deficiencies best explains this condition?

A Myophosphorylase

CORRECT

B Glucose-6-phosphatase

C Glycogen phosphorylase (hepatic)

D Hexokinase

RIGHT ANSWER

OK **A. Myophosphorylase**

WHY THIS IS RIGHT

McArdle disease (Glycogen Storage Disease Type V) is caused by deficiency of myophosphorylase, the muscle isoform of glycogen phosphorylase, which is required for breakdown of glycogen to glucose-1-phosphate in skeletal muscle.

- Inability to utilize muscle glycogen during exercise
- Leads to energy deficiency in muscle cells during exertion

Clinical features:

- Exercise intolerance (early fatigue)
- Muscle cramps and pain
- Myoglobinuria (dark urine after exercise)
- "Second-wind phenomenon" (symptoms improve after rest due to use of blood glucose)

Why other options are incorrect:

- Glucose-6-phosphatase: Deficient in Von Gierke disease (GSD I)
- Glycogen phosphorylase: General term; the specific deficient enzyme in muscle is myophosphorylase
- Hexokinase: Involved in glycolysis, not glycogen breakdown

QUESTION

Which of the following acts as an inhibitor of Complex IV (Cytochrome c oxidase) in the electron transport chain?

A Cyanide

CORRECT

B Carbon Dioxide

C Oligomycin

D Antimycin

RIGHT ANSWER

OK A. Cyanide

WHY THIS IS RIGHT

Complex IV (Cytochrome c oxidase) is the final enzyme of the electron transport chain (ETC) and is responsible for transferring electrons to oxygen, forming water.

Cyanide is a potent inhibitor of Complex IV. It binds to the ferric (Fe^{3+}) iron of cytochrome a_3 , blocking electron transfer to oxygen. This halts the entire ETC, leading to:

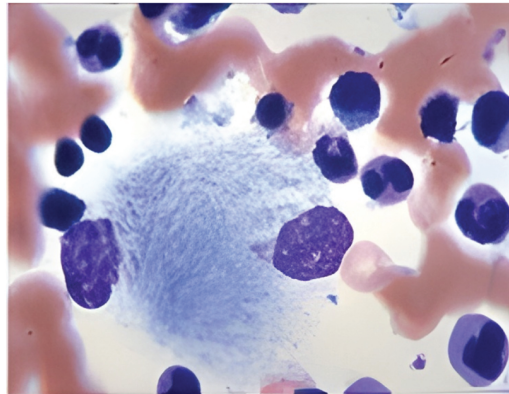
- Cessation of oxidative phosphorylation
- ↓ ATP production
- Cells switch to anaerobic glycolysis → lactic acidosis
- Causes histotoxic hypoxia (oxygen is present but cannot be utilized)

Why other options are incorrect:

- Carbon dioxide: Not an ETC inhibitor
- Oligomycin: Inhibits ATP synthase (Complex V)
- Antimycin A: Inhibits Complex III

QUESTION

The following histological image is seen in the patient who is complaining of growth failure and abdominal distension. Signs found were anemia, thrombocytopenia and bony pain. Which enzyme is deficient in this patient?



A Beta-Glucocerebrosidase

CORRECT

B Sphingomyelinase

C Hexosaminidase

D G-6-phosphatase

RIGHT ANSWER

A. Beta-Glucocerebrosidase

WHY THIS IS RIGHT

The histological image shows a Gaucher cell—a lipid-laden macrophage with a “crumpled tissue paper” cytoplasm, which is characteristic of Gaucher disease.

- Deficiency of β -glucocerebrosidase (β -glucosidase)
- Leads to accumulation of glucocerebroside in macrophages of the liver, spleen, and bone marrow

Clinical features explained:

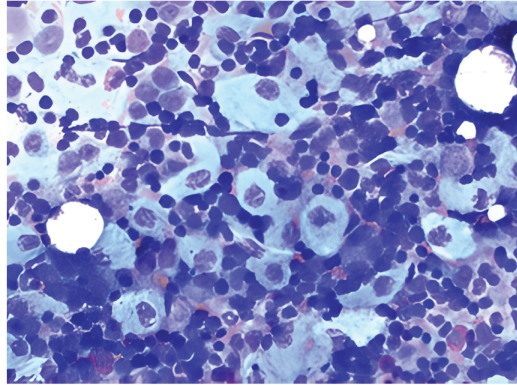
- Hepatosplenomegaly → abdominal distension
- Bone marrow infiltration → anemia + thrombocytopenia
- Bone involvement → bone pain and fractures
- Growth failure in children

Why other options are incorrect:

- Sphingomyelinase deficiency: Niemann-Pick disease (foam cells, neurodegeneration)
- Hexosaminidase deficiency: Tay-Sachs disease (cherry red spot, no organomegaly)
- G-6-phosphatase deficiency: Von Gierke disease (hypoglycemia, hepatomegaly)

QUESTION

An individual undergoes a bone marrow examination which shows the presence of the cells as shown in the image. Which of the following is the likely enzyme defect associated with this condition?



A Hexosaminidase

B Glucocerebrosidase

CORRECT

C Alpha 1,4 glucosidase

D Galactosidase

RIGHT ANSWER

B. Glucocerebrosidase

WHY THIS IS RIGHT

The bone marrow image shows Gaucher cells-large macrophages with abundant, wrinkled (“crumpled tissue paper”) cytoplasm, which is characteristic of Gaucher disease.

- Deficiency of glucocerebrosidase (β -glucosidase)
- Leads to accumulation of glucocerebroside in macrophages

Pathophysiology:

- Lipid-laden macrophages infiltrate bone marrow, liver, and spleen
- Causes marrow suppression -> anemia, thrombocytopenia
- Bone involvement -> bone pain, fractures
- Hepatosplenomegaly

Why other options are incorrect:

- Hexosaminidase: Tay-Sachs disease (neurodegeneration, cherry red spot)
- Alpha-1,4-glucosidase: Pompe disease (cardiomegaly, muscle weakness)
- Galactosidase: Fabry disease (angiokeratomas, neuropathy)

QUESTION

A child presents with mental retardation, bone pain, and inability to walk. On fundoscopy examination, a cherry red spot is seen. There is no organomegaly. What is the most likely diagnosis?

A Tay-Sachs disease

CORRECT

B Niemann-Pick disease

C Hurler syndrome

D Gaucher disease

RIGHT ANSWER

OK **A. Tay-Sachs disease**

WHY THIS IS RIGHT

A child with neurodegeneration (mental retardation), motor difficulty, and cherry red spot on fundoscopy without organomegaly is characteristic of Tay-Sachs disease.

- Deficiency of hexosaminidase A
- Leads to accumulation of GM2 ganglioside in neurons
- Lipid accumulation in neurons -> progressive neurodegeneration
- Retina appears pale due to storage -> fovea stands out as cherry red spot

Important differentiation:

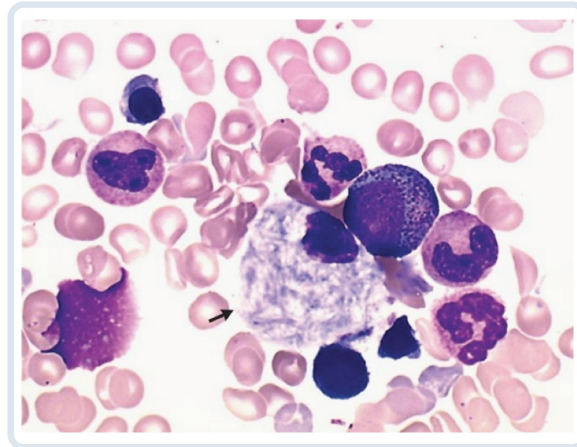
- Tay-Sachs: Cherry red spot without organomegaly
- Niemann-Pick: Cherry red spot with hepatosplenomegaly

Why other options are incorrect:

- Niemann-Pick disease: Has hepatosplenomegaly (foam cells)
- Hurler syndrome: Coarse facies, corneal clouding (no cherry red spot)
- Gaucher disease: Hepatosplenomegaly, bone pain, no cherry red spot

QUESTION

A child presents with bone pain and hepatosplenomegaly. A trephine biopsy and aspirate show the following finding. Which of the following is the most likely enzyme deficient in this condition?



A Hexosaminidase A

B. Glucocerebrosidase

CORRECT

C Sphingomyelinase

D α -1,4-Glucosidase

RIGHT ANSWER

B. Glucocerebrosidase

WHY THIS IS RIGHT

A child with bone pain, hepatosplenomegaly, and characteristic bone marrow findings (Gaucher cells) is diagnosed with Gaucher disease, a lysosomal storage disorder.

Underlying enzyme defect: Deficiency of glucocerebrosidase (β -glucosidase)

- Accumulation of glucocerebroside in macrophages
- These macrophages appear as Gaucher cells with "crumpled tissue paper" cytoplasm
- Infiltration of organs leads to:

- o Hepatosplenomegaly
- o Bone marrow suppression -> anemia, thrombocytopenia
- o Bone pain and fractures

Why other options are incorrect:

- Hexosaminidase A: Tay-Sachs (cherry red spot, no organomegaly)
- Sphingomyelinase: Niemann-Pick (foam cells + hepatosplenomegaly)
- α -1,4-glucosidase: Pompe disease (cardiomegaly, muscle weakness)

QUESTION

A chronic alcoholic patient is diagnosed with gouty arthritis. Mark the correct biochemical changes which will be seen in this patient?

- A** Decreased NADH/NAD⁺ ratio
- B** Increased urea and urate levels
- C** Increased urate and lactate levels
- D** Increased level of alkaline phosphatase

CORRECT

RIGHT ANSWER

- OK** **C. Increased urate and lactate levels**

WHY THIS IS RIGHT

In chronic alcoholism, metabolism of ethanol in the liver leads to an increased NADH/NAD⁺ ratio due to the actions of alcohol dehydrogenase and aldehyde dehydrogenase.

Metabolic consequences:

- ↑ NADH shifts pyruvate → lactate → causes lactic acidosis
- Lactate competes with uric acid for renal excretion
- ↓ Excretion of uric acid → hyperuricemia → gout

Why other options are incorrect:

- Decreased NADH/NAD⁺ ratio: Opposite; alcohol increases NADH
- Increased urea and urate: Urea is not the key change linked to gout here
- Increased alkaline phosphatase: Not specific to gout or alcohol metabolism

QUESTION

A 4-year-old child presents with easy fatiguability. The mother also complains that the child has increased hunger between meals, which is relieved after food. Liver examination revealed no glycogen. What enzyme is most likely deficient?

A Glycogen phosphorylase

B Glycogen synthase

CORRECT

C Debranching enzyme

D Glucose 6 phosphatase

RIGHT ANSWER

OK B. Glycogen synthase

WHY THIS IS RIGHT

Glycogen synthase deficiency (Glycogen Storage Disease Type 0) is characterized by an inability to synthesize glycogen, especially in the liver.

- Glycogen synthase catalyzes formation of glycogen from glucose
- Deficiency -> absent/markedly reduced glycogen stores in liver

Clinical features explained:

- Fasting hypoglycemia -> easy fatigability
- Increased hunger between meals (ketotic hypoglycemia)
- Symptoms improve after feeding
- No hepatomegaly (because glycogen is not stored)

Why other options are incorrect:

- Glycogen phosphorylase: Deficiency causes Hers disease (GSD VI) -> hepatomegaly
- Debranching enzyme: Causes Cori disease (GSD III) -> hepatomegaly + abnormal glycogen
- Glucose-6-phosphatase: Causes Von Gierke disease (GSD I) -> severe hypoglycemia + hepatomegaly

QUESTION

A 12-year-old boy is brought to the OPD as he is always fatigued and has muscle cramps after exercise and the blood sample collected soon after exercise showed low glucose and lactate level. He also has a history of one episode of hypoglycaemia while playing. He became normal after resting for a while. Which of the following diseases is he most likely to be suffering from?

A McArdle disease

CORRECT

B Hers disease

C Cori's disease

D Andersen disease

RIGHT ANSWER

OK **A. McArdle disease**

WHY THIS IS RIGHT

McArdle disease (Glycogen Storage Disease Type V) is caused by deficiency of myophosphorylase, the enzyme responsible for breakdown of glycogen in skeletal muscle.

- Muscle cannot convert glycogen → glucose-1-phosphate
- Leads to failure of glycolysis during exercise
- Hence, ↓ lactate production after exercise (key diagnostic clue)

Clinical features explained:

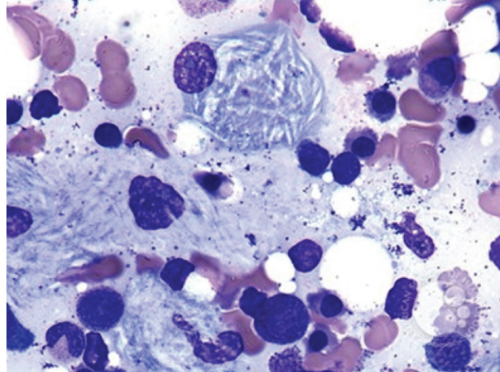
- Exercise intolerance & early fatigue
- Muscle cramps after exercise
- Low lactate levels post-exercise
- Second-wind phenomenon (symptoms improve after rest due to use of blood glucose)
- Occasional hypoglycemia during exertion

Why other options are incorrect:

- Hers disease (GSD VI): Liver glycogen phosphorylase deficiency → mild hypoglycemia + hepatomegaly
- Cori disease (GSD III): Debranching enzyme deficiency → hepatomegaly + abnormal glycogen
- Andersen disease (GSD IV): Branching enzyme deficiency → liver cirrhosis, not exercise intolerance

QUESTION

A crumpled tissue paper appearance and pancytopenia are seen in which enzyme defect?



A Alpha galactosidase

B Alpha glucosidase

C Beta galactosidase

D **Glucocerebrosidase**

CORRECT

RIGHT ANSWER

OK

D. Glucocerebrosidase

WHY THIS IS RIGHT

The "crumpled tissue paper" appearance of macrophages in bone marrow is characteristic of Gaucher disease, caused by deficiency of glucocerebrosidase.

- Deficiency of β -glucocerebrosidase
- Leads to accumulation of glucocerebroside in macrophages
- Lipid-laden macrophages (Gaucher cells) accumulate in:

- o Bone marrow
- o Liver
- o Spleen
- Causes pancytopenia due to marrow infiltration

Clinical features:

- Hepatosplenomegaly
- Pancytopenia (anemia, thrombocytopenia)
- Bone pain and fractures

Why other options are incorrect:

- Alpha-galactosidase: Fabry disease
- Alpha-glucosidase: Pompe disease
- Beta-galactosidase: GM1 gangliosidosis

QUESTION

A neonate was brought with complaints of poor feeding, vomiting, acidosis, and cataract. Urine Benedict's test was positive, while urine glucose was negative. The enzyme defective in this case is:

A Fructokinase

B Sucrase

C Galactose-1-phosphate uridyl transferase

CORRECT

D Lactase

RIGHT ANSWER

OK

C. Galactose-1-phosphate uridyl transferase

WHY THIS IS RIGHT

A neonate with poor feeding, vomiting, acidosis, cataracts, and positive Benedict's test (reducing sugar present) but negative glucose is classic for galactosemia.

Deficiency of galactose-1-phosphate uridyl transferase (GALT)

- Inability to convert galactose-1-phosphate → UDP-galactose
- Accumulation of:

o Galactose-1-phosphate (toxic to liver)

o Galactitol (via aldose reductase → causes cataracts)

Clinical features explained:

- Vomiting, poor feeding: After milk intake (lactose → glucose + galactose)
- Hepatic dysfunction → acidosis
- Cataracts: Due to galactitol accumulation
- Benedict's positive, glucose negative: Indicates non-glucose reducing sugar (galactose)

Why other options are incorrect:

- Fructokinase: Essential fructosuria (benign)
- Sucrase: Causes sucrose intolerance (no cataracts)
- Lactase: Causes lactose intolerance (diarrhea, no systemic toxicity)

QUESTION

| Which of the following are Methanol poisoning derivatives?

A Formic acid + Oxalic acid

B Formic acid + Lactic acid

CORRECT

C Glycolic acid and Oxalic acid

D Oxalic acid and Lactic acid

RIGHT ANSWER

OK B. Formic acid + Lactic acid

WHY THIS IS RIGHT

Methanol poisoning leads to formation of toxic metabolites through alcohol metabolism in the liver:
Methanol → Formaldehyde → Formic acid

Key toxic metabolite:

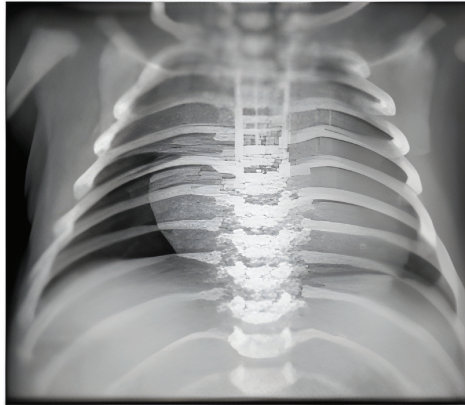
- Formic acid → causes metabolic acidosis and optic nerve toxicity (visual disturbances/blindness)
- Tissue hypoxia due to mitochondrial inhibition → ↑ lactic acid production

Why other options are incorrect:

- Oxalic acid: Seen in ethylene glycol poisoning (causes renal failure, calcium oxalate crystals)
- Glycolic acid: Also from ethylene glycol metabolism

QUESTION

A patient present with an X-ray showing cardiomegaly, along with symptoms of hypotonia, macroglossia, hepatomegaly, and "floppy baby" syndrome. What is the most likely diagnosis?



A Pompe's disease

CORRECT

B Ebstein anomaly

C Transposition of great arteries

D Von Gierke's disease

RIGHT ANSWER

OK **A. Pompe's disease**

WHY THIS IS RIGHT

The combination of cardiomegaly, hypotonia (floppy baby), macroglossia, and hepatomegaly is characteristic of Pompe disease (Glycogen Storage Disease Type II).

Deficiency of acid α -1,4-glucosidase (acid maltase)

- Impaired breakdown of glycogen in lysosomes
- Leads to glycogen accumulation in cardiac and skeletal muscle
- Causes massive cardiomegaly and muscle weakness

Clinical features explained:

- Cardiomegaly $\rightarrow$ heart failure
- Hypotonia $\rightarrow$ "floppy baby"
- Macroglossia
- Hepatomegaly

Why other options are incorrect:

- Ebstein anomaly: Congenital heart defect, not metabolic
- Transposition of great arteries: Cyanotic heart disease
- Von Gierke disease: Severe hypoglycemia + hepatomegaly (no cardiomegaly)

QUESTION

A patient is being treated with phenobarbitone. Which of the following is inhibited?

A Complex I -> coenzyme Q

CORRECT

B Complex II -> coenzyme Q

C Complex III -> Cyt. C

D Complex IV -> oxygen

RIGHT ANSWER

OK **A. Complex I -> coenzyme Q**

WHY THIS IS RIGHT

Phenobarbitone (barbiturates) can inhibit the electron transport chain at Complex I (NADH dehydrogenase), specifically blocking the transfer of electrons from NADH to coenzyme Q (ubiquinone).

- Inhibits electron flow from NADH -> CoQ
- Prevents proton pumping at Complex I
- Leads to ↓ ATP production via oxidative phosphorylation

Consequences:

- Cells shift toward anaerobic glycolysis
- May result in lactic acidosis in severe toxicity
- CNS depression due to reduced energy availability

Why other options are incorrect:

- Complex II -> CoQ: Inhibited by malonate
- Complex III -> Cytochrome C: Inhibited by antimycin A
- Complex IV -> O₂: Inhibited by cyanide, CO

QUESTION

A 5-year-old girl was washing her doll with shampoo containing rotenone. Her mother noticed her in an unconscious state. Which is inhibited by the above chemical?

A NADH dehydrogenase

CORRECT

B Succinate dehydrogenase

C Cytochrome C

D Cytochrome oxidase

RIGHT ANSWER

OK

A. NADH dehydrogenase

WHY THIS IS RIGHT

Rotenone is a toxin that inhibits Complex I (NADH dehydrogenase) of the electron transport chain (ETC) in mitochondria.

- Blocks transfer of electrons from NADH to coenzyme Q (ubiquinone)
- Prevents proton pumping across inner mitochondrial membrane
- Leads to ↓ ATP production (oxidative phosphorylation failure)

Consequences:

- Cells switch to anaerobic metabolism -> lactic acidosis
- High-energy tissues (brain) are affected first -> unconsciousness

Why other options are incorrect:

- Succinate dehydrogenase (Complex II): Inhibited by malonate
- Cytochrome C: Electron carrier, not directly inhibited here
- Cytochrome oxidase (Complex IV): Inhibited by cyanide and CO

QUESTION

A 6-month-old infant presents with protuberant abdomen, doll-like facies, and recurrent episodes of hypoglycemia. On examination, there is marked hepatomegaly. Laboratory evaluation suggests Von Gierke disease. What is the primary mechanism responsible for the enlarged liver in this condition?

- A** Excess deposition of glycogen within hepatocytes
- B** Accumulation of glucose-6-phosphate due to enzymatic block
- C** Increased levels of glucose-1-phosphate
- D** Defective glycogen primer (glycogenin) formation

CORRECT

RIGHT ANSWER

- OK** **A. Excess deposition of glycogen within hepatocytes**

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$1a” here and does not include a written explanation for this item.

QUESTION

| Which of the following activate glycogen phosphorylase?

A Ca²⁺

CORRECT

B Glucose 6-Phosphate

C Glucose

D High ATP

RIGHT ANSWER

OK A. Ca²⁺

WHY THIS IS RIGHT

No explanation was provided in the source quiz.

QUESTION

Congenital lactic acidosis is due to the defect of

- A** Transketolase
- B** Branched chain ketoacid dehydrogenase
- C** Isocitrate dehydrogenase
- D** **Pyruvate dehydrogenase**

CORRECT

RIGHT ANSWER

- OK** **D. Pyruvate dehydrogenase**

WHY THIS IS RIGHT

Congenital lactic acidosis commonly results from a deficiency of the pyruvate dehydrogenase (PDH) complex, which converts pyruvate to acetyl-CoA, linking glycolysis to the TCA cycle in mitochondria. When PDH is defective, pyruvate cannot enter the TCA cycle and is instead converted to lactate by lactate dehydrogenase, leading to accumulation of lactic acid and metabolic acidosis. Patients often present with neurological deficits, developmental delay, hypotonia, and persistent lactic acidosis. Thus, pyruvate dehydrogenase deficiency impairs aerobic glucose metabolism and causes congenital lactic acidosis.

Why not others:

- Transketolase: Enzyme of the HMP shunt (pentose phosphate pathway), not involved in pyruvate metabolism.
- Branched-chain ketoacid dehydrogenase: Deficiency causes Maple syrup urine disease, not lactic acidosis.
- Isocitrate dehydrogenase: TCA cycle enzyme; defects are mainly associated with certain tumors, not congenital lactic acidosis.

QUESTION

Match the following disorders with their respective clinical features or lab findings.

Disorder	Clinical Feature or Lab Findings
1. Cori's disease	A. Branched structure in liver biopsy
2. McArdle's disease	B. Accumulation of glycogen in lysosome
3. Pompe's disease	C. Exercise intolerance
4. Von Gierke's disease	D. Hypoglycaemia with lactic acidosis

A 1 -> B, 2 -> C, 3 -> A, 4 -> D

B 1 -> C, 2 -> B, 3 -> A, 4 -> D

C 1 -> A, 2 -> C, 3 -> B, 4 -> D

CORRECT

D 1 -> A, 2 -> C, 3 -> D, 4 -> B

RIGHT ANSWER

OK C. 1 -> A, 2 -> C, 3 -> B, 4 -> D

WHY THIS IS RIGHT

No explanation was provided in the source quiz.

Disorder	Finding	Key point
1. Cori's disease	A. Branched structure in liver biopsy	Debranching enzyme deficiency → short outer chains
2. McArdle's disease	C. Exercise intolerance	No rise in lactate with exercise; "second-wind phenomenon"
3. Pompe's disease	B. Glycogen in lysosomes	Only glycogen storage disease that is lysosomal and causes cardiomyopathy
4. Von Gierke's disease	D. Hypoglycaemia + lactic acidosis	G6-phosphatase deficiency → hepatomegaly, doll-face appearance

QUESTION

| Which of the following is not a cytochrome C oxidase inhibitor?

A Carbon monoxide

B Nitric oxide

CORRECT

C Hydrogen sulfide

D Cyanide

RIGHT ANSWER

OK B. Nitric oxide

WHY THIS IS RIGHT

Cytochrome c oxidase (Complex IV) is the final enzyme of the mitochondrial electron transport chain that transfers electrons from cytochrome c to oxygen, forming water and enabling oxidative phosphorylation and ATP production.

Several toxic substances inhibit this enzyme by binding to the heme-iron center, thereby blocking electron transfer to oxygen and halting cellular respiration. Important inhibitors include:

- Cyanide
- Carbon monoxide
- Hydrogen sulfide
- Azide

Inhibition leads to histotoxic hypoxia, where oxygen is present but cells cannot utilize it for ATP generation.

Nitric oxide (NO), although it can interact with mitochondrial enzymes in signaling pathways, is not classically considered a cytochrome c oxidase inhibitor.

QUESTION

A patient presents with hepatosplenomegaly and hypoglycemia. After a meal, glucagon administration increases blood glucose levels. However, after an overnight fast, glucagon administration does not improve blood glucose levels. Liver biopsy reveals increased abnormal glycogen accumulation. Which enzyme deficiency is most likely?

A Muscle phosphorylase

B Glycogen phosphorylase

C Debranching enzyme

CORRECT

D Glucose-6-phosphatase

RIGHT ANSWER

OK C. Debranching enzyme

WHY THIS IS RIGHT

Debranching enzyme deficiency (Glycogen Storage Disease type III - Cori disease) leads to accumulation of abnormally structured glycogen with short outer branches (limit dextrin) in the liver.

Key Clinical Features

- Hepatomegaly and hypoglycemia
- Abnormal glycogen accumulation in liver
- Partial ability to mobilize glycogen

Key Diagnostic Clue

- After a meal: glucagon increases glucose because some glycogen breakdown is still possible.
- After prolonged fasting: glucagon cannot increase glucose, as the remaining glycogen cannot be further degraded due to lack of debranching enzyme.

| Disease | Enzyme Defect | Key Feature |

| ----- | ----- | ----- |

| Von Gierke (Type I) | Glucose-6-phosphatase | Severe fasting hypoglycemia, lactic acidosis |

| Cori disease (Type III) | Debranching enzyme | Limit dextrin accumulation, hepatomegaly |

| McArdle disease (Type V) | Muscle phosphorylase | Exercise intolerance, myoglobinuria |

QUESTION

Carbon monoxide and cyanide cause death primarily due to their interference with which of the following cellular processes?

A Myoglobin

B Cytochrome oxidase

CORRECT

C Hemoglobin

D NADH Dehydrogenase

RIGHT ANSWER

B. Cytochrome oxidase

WHY THIS IS RIGHT

Carbon monoxide and cyanide cause cellular hypoxia by inhibiting the final step of the mitochondrial electron transport chain.

- Cyanide binds to cytochrome c oxidase (Complex IV) in mitochondria.
- This blocks the transfer of electrons to oxygen, preventing formation of water.
- As a result, oxidative phosphorylation stops, ATP production ceases, and cells develop histotoxic hypoxia (oxygen is present but cannot be utilized).

Carbon monoxide (CO) contributes to hypoxia by two mechanisms:

1. High affinity binding to hemoglobin -> formation of carboxyhemoglobin -> reduced oxygen delivery.
2. Binding to mitochondrial cytochrome oxidase -> inhibition of cellular respiration\.

QUESTION

A patient presents with hepatosplenomegaly and a cherry-red spot on the macula. Which enzyme deficiency is most likely responsible for this clinical picture?

A Sphingomyelinase

CORRECT

B Hexosaminidase A

C Glucose 6 phosphatase

D Galactocerebrosidase

RIGHT ANSWER

OK A. Sphingomyelinase

WHY THIS IS RIGHT

Lysosomal Storage Diseases and Associated Enzyme Deficiencies

Disease	Enzyme Deficiency
Tay-Sachs disease	Hexosaminidase A
Sandhoff's disease	Hexosaminidase A and B
Niemann-Pick disease	Sphingomyelinase
Metachromatic leukodystrophy	Arylsulfatase A
Krabbe disease	B-Galactosidase
Gaucher's disease	B-Glucosidase
Fabry's disease	α -Galactosidase
Farber's disease	Ceramidase

QUESTION

| The phenomenon of cancer cells consuming more glucose is due to?

A Lactic acid is produced despite presence of oxygen

CORRECT

B Enhanced conversion of glucose to pyruvate

C Increased pyruvate to acetyl CoA (High NADH/NAD ratio)

D Absence of Oxygen

RIGHT ANSWER

OK **A. Lactic acid is produced despite presence of oxygen**

WHY THIS IS RIGHT

Cancer cells preferentially utilize aerobic glycolysis, a phenomenon known as the Warburg effect. Even in the presence of adequate oxygen, cancer cells convert glucose to lactate instead of fully oxidizing pyruvate in mitochondria.

Consequences

- Increased glucose uptake
- Excess lactate production
- Provides biosynthetic intermediates needed for rapid cell proliferation.

QUESTION

A blood sample kept in vitro for metabolic analysis shows reduced glucose utilization along with a paradoxical increase in 2,3-BPG levels within red blood cells. The patient also has features suggestive of a chronic hemolytic state. Which enzyme deficiency best explains these findings?

- A** Hexokinase
- B** Pyruvate kinase
- C** Glucose-6-phosphate dehydrogenase

D Phosphoglycerate kinase

CORRECT

RIGHT ANSWER

OK D. Phosphoglycerate kinase

WHY THIS IS RIGHT

Phosphoglycerate kinase (PGK) deficiency blocks the glycolytic step converting 1,3-bisphosphoglycerate -> 3-phosphoglycerate, reducing ATP generation in red blood cells.

- Accumulation of upstream intermediates
- Increased diversion through the Rapoport-Luebering shunt
- ↑ 2,3-BPG levels
- Reduced glycolytic flux -> decreased glucose utilization in vitro

Elevated 2,3-BPG decreases hemoglobin's oxygen affinity, facilitating oxygen release to tissues but reflecting defective glycolysis in RBCs.

QUESTION

True statements about the HMP shunt:

1. Non-oxidative phase is reversible
2. End products are ribose-5-P, O_2 and NADH
3. No ATP produced in oxidative phase
4. Transketolase requires thiamine

A 1, 3 and 4

CORRECT

B 1 and 2

C 2, 3 and 4

D 1 and 4

RIGHT ANSWER

OK A. 1, 3 and 4

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$1b” here and does not include a written explanation for this item.

QUESTION

A 6-year-old child is brought with progressive abdominal distension. On examination, he has massive hepatomegaly and a doll-like facies with chubby cheeks. There is a history of recurrent episodes of irritability and sweating during fasting, which improve after feeding. The most likely metabolic abnormality in this child is:

- A** Fasting hypoglycemia
- B** Fasting hypoketosis
- C** Decreased hepatic glycogen stores
- D** Exercise-induced muscle cramps

CORRECT

RIGHT ANSWER

- OK** **A. Fasting hypoglycemia**

WHY THIS IS RIGHT

The presentation describes Glycogen Storage Disease Type I (Von Gierke disease) caused by deficiency of glucose-6-phosphatase.

Glucose-6-phosphatase is required to convert glucose-6-phosphate → free glucose in the liver and kidney during:

- Glycogenolysis
- Gluconeogenesis

Typical clinical features:

- Hepatomegaly due to glycogen accumulation
- Doll-like face (fat cheeks)
- Severe fasting hypoglycemia
- Lactic acidosis
- Hyperuricemia
- Hyperlipidemia

Why other options are incorrect:

- Fasting hypoketosis → seen in fatty acid oxidation defects, not Von Gierke disease
- Decreased hepatocellular glycogen → actually glycogen increases in the liver
- Muscle cramps → more typical of McArdle disease (GSD V)

QUESTION

A child is diagnosed with Gaucher's disease. Which statements are true?

1. Deficiency of sphingomyelinase
2. Deficiency of β -glucocerebrosidase
3. Presence of foam cells
4. Crumpled tissue-paper cytoplasm

A 2 and 4

CORRECT

B 1 and 3

C 1, 2 and 4

D 2, 3 and 4

RIGHT ANSWER

OK **A. 2 and 4**

WHY THIS IS RIGHT

Gaucher disease is a lysosomal storage disorder caused by deficiency of the enzyme β -glucocerebrosidase, leading to accumulation of glucocerebroside in macrophages.

- Enzyme deficiency: β -glucocerebrosidase -> impaired breakdown of glucocerebroside
- Affected cells: macrophages in liver, spleen, bone marrow
- Characteristic histology: macrophages with "crumpled tissue-paper" cytoplasm due to accumulated lipid

These macrophages are called Gaucher cells.

Why other statements are incorrect:

- Deficiency of sphingomyelinase -> seen in Niemann-Pick disease, not Gaucher disease.
- Foam cells -> typically described in Niemann-Pick disease due to sphingomyelin accumulation.

QUESTION

Which intermediate of the TCA cycle participates in fatty acid synthesis?

- A Malate
- B Fumarate
- C Succinate
- D Citrate**

CORRECT

RIGHT ANSWER

D. Citrate

WHY THIS IS RIGHT

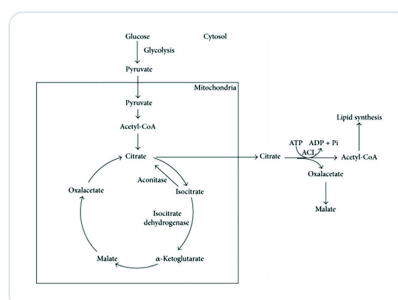
Citrate is the TCA cycle intermediate that links carbohydrate metabolism with fatty acid synthesis.

Role of citrate in fatty acid synthesis

- In the mitochondria, acetyl-CoA enters the TCA cycle and combines with oxaloacetate to form citrate.
- When cellular energy is abundant, citrate is transported from the mitochondria to the cytosol.
- In the cytosol, ATP-citrate lyase cleaves citrate into:
 - Acetyl-CoA
 - Oxaloacetate

The acetyl-CoA produced in the cytosol is the precursor for fatty acid synthesis.

Citrate also Activates acetyl-CoA carboxylase, the rate-limiting enzyme of fatty acid synthesis.



QUESTION

A 12-month-old male baby presents with vomiting, lethargy on consuming fruit juice. Which of the following can give a positive reaction in this patient's condition that is most diagnostic?

A Benedict's test

B Seliwanoff's reagent

CORRECT

C Sakaguchi's reagent

D Millon's reagent

RIGHT ANSWER

OK **B. Seliwanoff's reagent**

WHY THIS IS RIGHT

The patient has hereditary fructose intolerance (HFI), suggested by vomiting and lethargy after consuming fruit juice, which is high in fructose. Seliwanoff's test specifically detects ketose sugars like fructose by reacting with them to produce a red color. This is the most diagnostic chemical reaction for detecting fructose in this context.

Why the others are incorrect:

- A. Benedict's test: Detects reducing sugars (glucose, fructose, galactose), but is less specific for fructose compared to Seliwanoff's reagent.
- C. Sakaguchi's reagent: Detects arginine, unrelated to sugar metabolism.
- D. Millon's reagent: Detects phenolic compounds like tyrosine, not fructose.

QUESTION

A 6-month-old girl is brought to the OPD by her mother for a check-up appointment. The mother states, "My baby doesn't seem to be growing much despite feeding as often as my previous children I'm worried that something is wrong with her." Height and weight are below the 10th percentile. Physical examination shows hepatomegaly and hypotonia. Laboratory results show hypoglycemia and ketoacidosis. Liver biopsy shows hepatic fibrosis without fat accumulation. Further analysis reveals excessive amounts of limit dextrins within the cytosol of the hepatocytes. Which of the following enzymes is most likely deficient in this patient?

- A** Acid alpha-glucosidase
- B** Glucose-6-phosphatase
- C** Glycogen debrancher enzyme
- D** Liver glycogen phosphorylase

CORRECT

RIGHT ANSWER

- OK** C. Glycogen debrancher enzyme

WHY THIS IS RIGHT

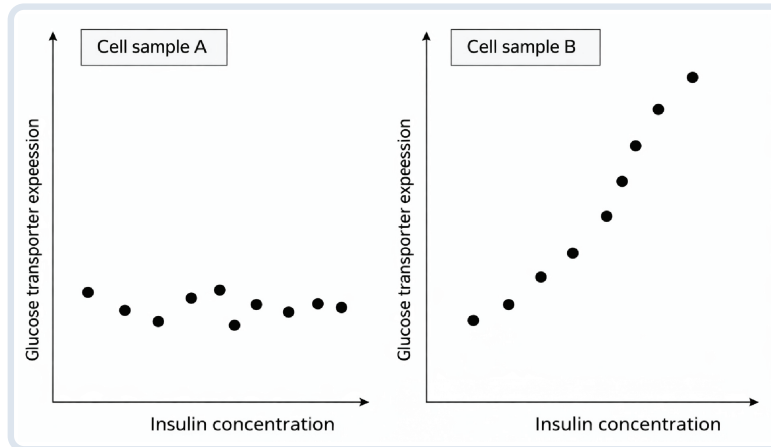
This 6-month-old has Cori disease (glycogen storage disease type III), suggested by hypoglycemia, hepatomegaly, hypotonia, growth delay, and ketoacidosis. The liver biopsy shows excess limit dextrins-short outer branches of glycogen-because the glycogen debranching enzyme is deficient. Debranching enzyme normally allows complete glycogen breakdown by removing α -1,6-linked branches.

Why the others are incorrect:

- A. Acid α -glucosidase: Deficiency causes Pompe disease, leading to lysosomal glycogen accumulation and cardiomyopathy, not cytosolic limit dextrins.
- B. Glucose-6-phosphatase: Deficiency causes Von Gierke disease, with severe fasting hypoglycemia, hepatomegaly, and lactic acidosis; glycogen is structurally normal.
- D. Liver glycogen phosphorylase: Deficiency causes GSD type VI (Hers disease); glycogen accumulates, but limit dextrins do not.

QUESTION

Researchers are investigating the relationship between glucose transport proteins and insulin concentration in different types of tissues. Experiments in which cells are taken from various tissue samples and exposed to increasing concentrations of insulin are performed. The number of surface glucose transporters are then measured. The results from 2 cell samples are plotted on the graphs below. Which of the following cell types are most likely represented in cell samples A and B, respectively?



- A** A-Neurons, B-Hepatocytes
- B** A-Intestinal epithelial, B-Pancreatic beta cells
- C** A-Renal tubular cells, B- Hepatocytes
- D** A-Renal tubular cells, B-Skeletal myocytes

CORRECT

RIGHT ANSWER

- OK** D. A-Renal tubular cells, B-Skeletal myocytes

WHY THIS IS RIGHT

- Renal tubular cells reabsorb glucose via SGLTs (sodium-glucose cotransporters), which are largely insulin-independent, so their surface transporter number doesn't change much with insulin (Cell A).
- Skeletal myocytes express GLUT4, which is insulin-dependent. Increasing insulin causes more GLUT4 to translocate to the membrane, increasing glucose uptake (Cell B), matching the observed graph trend.

Why the others are incorrect:

A. Neurons & hepatocytes: Neurons are insulin-independent, but hepatocytes also uptake glucose largely insulin-independently (GLUT2), so they wouldn't show a strong insulin-response curve.

B. Intestinal epithelial & pancreatic beta cells: These cells either do not respond strongly to insulin (intestinal) or sense glucose rather than transport it in response to insulin (beta cells).

QUESTION

A diabetic man takes his insulin bolus and then takes his meal. This would cause activation of all of the following enzymes, except:

A Phosphofructokinase

B Phosphoenolpyruvate carboxykinase

CORRECT

C Glycogen synthase

D Acetyl CoA carboxylase

RIGHT ANSWER

OK B. Phosphoenolpyruvate carboxykinase

WHY THIS IS RIGHT

After insulin bolus and a meal, the body is in a fed state, promoting anabolic pathways:

- Phosphofructokinase: Activated by insulin -> stimulates glycolysis.
- Glycogen synthase: Activated by insulin -> promotes glycogen synthesis.
- Acetyl-CoA carboxylase: Activated by insulin -> promotes fatty acid synthesis.
- PEPCK: A key gluconeogenic enzyme, which is inhibited by insulin because gluconeogenesis is suppressed in the fed state.

Why other options are incorrect:

- PFK, glycogen synthase, and ACC are all upregulated/activated by insulin to store glucose and energy.
- Only PEPCK is downregulated to prevent glucose production when exogenous glucose is available.

QUESTION

Identify the correct statements

1. Urinary levels of 3-methylhistidine are high in patients with Wilson disease.
2. Hyaluronic acid is the longest GAG
3. Keratan sulfate is the only GAG without uronic acid
4. Heparan sulfate is a major component of the glomerular basement membrane (GBM).

A 2, 4

B 1, 3, 4

C 2, 3, 4

CORRECT

D 1, 4

RIGHT ANSWER

OK C. 2, 3, 4

WHY THIS IS RIGHT

- Hyaluronic acid is the longest GAG (2): True; it is a non-sulfated glycosaminoglycan with very high molecular weight.
- Keratan sulfate is the only GAG without uronic acid (3): True; its repeating units contain galactose and N-acetylglucosamine instead of uronic acids.
- Heparan sulfate is a major component of GBM (4): True; it contributes to the negative charge and selective filtration in the glomerular basement membrane.

Why the other option is incorrect:

Urinary 3-methylhistidine in Wilson disease (1): False; elevated urinary 3-methylhistidine indicates muscle protein breakdown, not directly related to Wilson disease.

QUESTION

Choose the correct statements about glucokinase.

1. Located in β cells of the pancreas.
2. Has a lower affinity for glucose.
3. Is induced by insulin.
4. Inhibited by glucose-6-phosphate
5. Mutations can lead to MODY.

A 2, 4, 5

B 3, 4, 5

C 1, 2, 3, 5

CORRECT

D 1, 2, 3, 4

RIGHT ANSWER

OK C. 1, 2, 3, 5

WHY THIS IS RIGHT

Located in β cells of the pancreas (1): True; glucokinase acts as a glucose sensor in pancreatic β cells and also in hepatocytes.

Has a lower affinity for glucose (2): True; it has a high K_m (~ 10 mM), allowing it to respond to high glucose concentrations.

Is induced by insulin (3): True; insulin upregulates hepatic glucokinase expression.

Inhibited by glucose-6-phosphate: False; unlike hexokinase, glucokinase is not inhibited by glucose-6-phosphate.

Mutations can lead to MODY (5): True; GCK mutations cause MODY2, a mild form of diabetes.

Why other options are incorrect:

Option 2,4,5 includes inhibition by G6P, which is false.

Option 3,4,5 and 1,2,3,4 incorrectly include G6P inhibition.

QUESTION

Match the following disorders with their respective clinical features or lab findings.

Disorder	Clinical Feature or Lab Findings
1. Cori's disease	A. Branched structure in liver biopsy
2. McArdle's disease	B. Accumulation of glycogen in lysosome
3. Pompe's disease	C. Exercise intolerance
4. Von Gierke's disease	D. Hypoglycemia with lactic acidosis

A 1 -> B, 2 -> C, 3 -> A, 4 -> D

B 1 -> C, 2 -> B, 3 -> A, 4 -> D

C 1 -> A, 2 -> C, 3 -> B, 4 -> D

CORRECT

D 1 -> A, 2 -> C, 3 -> D, 4 -> B

RIGHT ANSWER

OK C. 1 -> A, 2 -> C, 3 -> B, 4 -> D

WHY THIS IS RIGHT

Cori disease (1 -> A): Caused by debranching enzyme deficiency, leading to abnormally branched glycogen with short outer chains seen on liver biopsy.

McArdle disease (2 -> C): Muscle glycogen phosphorylase deficiency causes exercise intolerance, muscle cramps, and myoglobinuria.

Pompe disease (3 -> B): Lysosomal acid α -glucosidase deficiency results in glycogen accumulation in lysosomes, especially in muscle and heart.

Von Gierke disease (4 -> D): Glucose-6-phosphatase deficiency causes severe hypoglycemia with lactic acidosis due to impaired glucose release.

QUESTION

An 8-month-old infant is brought to the pediatrician for evaluation of multiple episodes of seizures and hypoglycemia. A liver biopsy demonstrates hepatocytes with large glycogen stores. Biochemical analysis reveals glycogen with short outer chains. Which of the following is the likely diagnosis?

A Cori disease

CORRECT

B Anderson disease

C Von-Gierke disease

D Pompe disease

RIGHT ANSWER

OK A. Cori disease

WHY THIS IS RIGHT

Cori disease (Glycogen storage disease type III) is caused by deficiency of the debranching enzyme, leading to accumulation of abnormally structured glycogen with short outer chains. Affected infants present with hepatomegaly, hypoglycemia, seizures, and excess glycogen in hepatocytes. Unlike Von Gierke disease, gluconeogenesis is intact, so hypoglycemia is less severe.

Why other options are incorrect:

B. Anderson disease: Defect in branching enzyme -> long, poorly branched glycogen causing liver cirrhosis.

C. Von-Gierke disease: Glucose-6-phosphatase deficiency; glycogen structure is normal.

D. Pompe disease: Lysosomal acid maltase deficiency with cardiomegaly, not hypoglycemia.

QUESTION

Which of the following statements correctly differentiates liver glycogen phosphorylase (LGP) and muscle glycogen phosphorylase (MGP)?

CORRECT

- A** LGP is activated by glucagon, while MGP is activated by epinephrine.
- B** LGP releases glucose-6-phosphate into the bloodstream, while MGP releases free glucose into the muscle.
- C** LGP activity is regulated by AMP, while MGP is regulated by glucagon and epinephrine.
- D** MGP is highly active during fasting compared to LGP.

RIGHT ANSWER

OK

A. LGP is activated by glucagon, while MGP is activated by epinephrine.

WHY THIS IS RIGHT

Liver glycogen phosphorylase (LGP) responds to glucagon and epinephrine to maintain blood glucose during fasting and stress. Muscle glycogen phosphorylase (MGP) is activated mainly by epinephrine, AMP, and Ca^{2+} during exercise to meet local energy needs; it does not respond to glucagon.

Why other options are incorrect:

B. LGP releases glucose-6-phosphate into the bloodstream, while MGP releases free glucose into the muscle: Liver releases free glucose (via glucose-6-phosphatase), while muscle releases glucose-6-phosphate, not vice versa.

C. LGP activity is regulated by AMP, while MGP is regulated by glucagon and epinephrine: Regulation is reversed-AMP regulates MGP, not LGP.

D. MGP is highly active during fasting compared to LGP: LGP is more active during fasting; MGP functions during muscle activity, not fasting.

QUESTION

A 6-month-old infant is brought with lethargy, vomiting, and poor feeding after a period of prolonged fasting. Labs show hypoglycemia with low ketone bodies (hypoketosis), suggesting a defect in fat metabolism. Which of the following is the most likely disorder?

- A** Hers disease
- B** Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
- C** Hereditary fructose intolerance
- D** Glycogen storage disease type I (Von Gierke disease)

CORRECT

RIGHT ANSWER

- OK** **B. Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency**

WHY THIS IS RIGHT

Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency is a fatty acid β -oxidation disorder in which the body cannot properly break down medium-chain fatty acids in mitochondria to generate energy during fasting.

In MCAD deficiency, impaired β -oxidation results in:

- Decreased energy production
- Reduced ketone body formation (hypoketosis)
- Hypoglycemia during fasting

This leads to symptoms such as lethargy, vomiting, poor feeding, and hypoketotic hypoglycemia, especially after prolonged fasting or illness.

Why other options are incorrect:

- Hers disease (GSD VI): Liver glycogen phosphorylase deficiency causing mild hypoglycemia and hepatomegaly, not hypoketotic hypoglycemia.
- Hereditary fructose intolerance: Presents after fructose ingestion with hypoglycemia and vomiting.
- Von Gierke disease (GSD I): Causes severe fasting hypoglycemia with hepatomegaly and lactic acidosis, but ketone production is not characteristically absent.

QUESTION

A patient with family history presents with tendon xanthomas. Laboratory investigations reveal elevated cholesterol with normal triglyceride levels. What is the most likely diagnosis?

- A** Familial hyperlipidemia
- B** Familial dysbetalipoproteinemia
- C** Familial hypertriglyceridemia
- D** **Familial hypercholesterolemia**

CORRECT

RIGHT ANSWER

- OK** **D. Familial hypercholesterolemia**

WHY THIS IS RIGHT

Familial hypercholesterolemia (FH) is an autosomal dominant disorder characterized by defective or absent LDL receptors, leading to impaired clearance of low-density lipoprotein (LDL) cholesterol from the blood. This results in marked elevation of LDL cholesterol with normal triglyceride levels.

The excess cholesterol gets deposited in tissues, producing characteristic findings such as:

- Tendon xanthomas (commonly in Achilles tendon and extensor tendons)
- Xanthelasma
- Premature atherosclerosis and early coronary artery disease

Why other options are incorrect:

- Familial hyperlipidemia: A nonspecific term and not a defined disorder.
- Familial dysbetalipoproteinemia: Characterized by elevation of both cholesterol and triglycerides with palmar xanthomas.
- Familial hypertriglyceridemia: Mainly shows elevated triglycerides, not isolated high cholesterol.

QUESTION

Fatty acid transport through mitochondrial membrane is via?

A Carnitine

CORRECT

B Acyl carrier protein

C Cholesterol transporter

D LCAT

RIGHT ANSWER

OK

A. Carnitine

WHY THIS IS RIGHT

Long-chain fatty acids cannot directly cross the inner mitochondrial membrane. Their transport into the mitochondrial matrix for β -oxidation requires the carnitine shuttle system.

Mechanism (Carnitine shuttle):

1. Fatty acids are activated to fatty acyl-CoA in the cytosol.
2. Carnitine palmitoyltransferase I (CPT-I) converts acyl-CoA to acyl-carnitine (outer mitochondrial membrane).
3. Acyl-carnitine is transported across the inner membrane via a translocase.
4. CPT-II reconverts acyl-carnitine to acyl-CoA inside the mitochondria for β -oxidation.

Defects in the carnitine shuttle (e.g., CPT deficiency) lead to impaired fatty acid oxidation $\rightarrow$ hypoketotic hypoglycemia and muscle weakness.

Why other options are incorrect:

- Acyl carrier protein: Involved in fatty acid synthesis, not transport.
- Cholesterol transporter: Not related to fatty acid mitochondrial entry.
- LCAT (Lecithin-cholesterol acyltransferase): Involved in cholesterol metabolism (HDL function).

QUESTION

A patient has multiple tendon xanthomas. Serum cholesterol (398 mg/dL) and LDL 220 mg/dL were found to be raised. What is the defect?

A Lipoprotein lipase deficiency

B LDL receptors defect

CORRECT

C Apo E defect

D LCAT deficiency

RIGHT ANSWER

B. LDL receptors defect

WHY THIS IS RIGHT

The presence of tendon xanthomas with markedly elevated total cholesterol and LDL is characteristic of familial hypercholesterolemia (Type IIa).

- Mutation in LDL receptor -> impaired uptake of LDL by liver
- Leads to accumulation of LDL in plasma
- Normal triglycerides

Pathophysiology:

- Excess LDL deposits cholesterol in tissues -> tendon xanthomas
- Promotes atherosclerosis -> premature coronary artery disease

Why other options are incorrect:

- Lipoprotein lipase deficiency: ↑ triglycerides (chylomicrons), not LDL
- Apo E defect: Dysbetalipoproteinemia (↑ cholesterol + TG, palmar xanthomas)
- LCAT deficiency: HDL metabolism defect (fish-eye disease), not tendon xanthomas

QUESTION

A patient came to the emergency room with severe pain in the abdomen. Serum triglyceride level was 1500 mg/dL. Identify the defect?

- A Apo B 48
- B Apo B 100
- C Apo CII**
- D LDL receptor

CORRECT

RIGHT ANSWER

C. Apo CII

WHY THIS IS RIGHT

Very high triglycerides (>1000 mg/dL) with acute abdominal pain suggests hypertriglyceridemia-induced pancreatitis, classically seen in familial chylomicronemia (Type I hyperlipoproteinemia).

- Deficiency of Apolipoprotein C-II (Apo C-II)
- Apo C-II is a cofactor for lipoprotein lipase (LPL)

Pathophysiology:

- Without Apo C-II -> LPL cannot be activated
- Impaired breakdown of chylomicrons -> massive accumulation of triglycerides
- Leads to pancreatitis, eruptive xanthomas, lipemia retinalis

Why other options are incorrect:

- Apo B-48: Needed for chylomicron formation (defect -> abetalipoproteinemia)
- Apo B-100: Required for LDL/VLDL metabolism (familial hypercholesterolemia-like states)
- LDL receptor: Causes ↑ LDL (Type IIa), not severe triglyceridemia

QUESTION

A patient presents with xanthomas on the Achilles tendon. Which of the following is the diagnosis?

- A Tangier's disease
- B Familial hyperchylomicronemia
- C Familial hypercholesterolemia**
- D Familial Dysbetalipoproteinemia

CORRECT

RIGHT ANSWER

- OK C. Familial hypercholesterolemia**

WHY THIS IS RIGHT

Tendon xanthomas (classically over the Achilles tendon) are a hallmark of familial hypercholesterolemia (Type II hyperlipoproteinemia). This condition is caused by a defect in LDL receptors, leading to markedly elevated LDL cholesterol levels and deposition of cholesterol in tendons and skin.

It is associated with premature atherosclerosis and early coronary artery disease.

Key differentiation:

- Familial hypercholesterolemia: Tendon xanthomas
- Familial dysbetalipoproteinemia: Palmar xanthomas
- Hyperchylomicronemia: Eruptive xanthomas
- Tangier disease: Low HDL, orange tonsils

QUESTION

Patient with high triglycerides esterified with long chain fatty acid (LCFA) presents with fatigue and biopsy of muscle shows fat vacuoles. Which of the following is the diagnosis?

A Carnitine deficiency

CORRECT

B FA synthase defect

C LPL defect

D LDL receptor defect

RIGHT ANSWER

OK

A. Carnitine deficiency

WHY THIS IS RIGHT

Carnitine deficiency is a disorder of fatty acid oxidation in which long-chain fatty acids (LCFA) cannot be transported into mitochondria for β -oxidation. Carnitine is required for the carnitine shuttle

- LCFA cannot be utilized -> accumulate as triglycerides in muscle
- Leads to lipid (fat) vacuoles in muscle biopsy
- $\downarrow$ energy production -> especially during fasting/exercise

Clinical features:

- Muscle weakness & fatigue
- Hypoketotic hypoglycemia
- Myopathy with lipid accumulation

Why other options are incorrect:

- Fatty acid synthase defect: Affects synthesis, not oxidation
- Lipoprotein lipase (LPL) defect: Causes $\uparrow$ triglycerides in plasma (chylomicronemia), not muscle fat vacuoles
- LDL receptor defect: Causes hypercholesterolemia, not fatty acid oxidation defect

QUESTION

A patient presents with seizures, a cherry-red spot on the macula, and no hepatosplenomegaly. Laboratory investigations reveal a deficiency of hexosaminidase. Which compound is accumulated in this patient?

A Galactosylceramide

B GM2 ganglioside

CORRECT

C Sphingomyelin

D GM1 ganglioside

RIGHT ANSWER

OK B. GM2 ganglioside

WHY THIS IS RIGHT

The clinical triad of seizures, neurodegeneration, cherry-red spot on macula, and absence of hepatosplenomegaly is characteristic of Tay-Sachs disease.

- Deficiency of hexosaminidase A
- Leads to accumulation of GM2 ganglioside in neurons

Pathophysiology:

- GM2 accumulates in lysosomes of neurons -> progressive neurodegeneration
- Retina becomes pale due to lipid accumulation, while the fovea appears as a cherry-red spot
- No organomegaly (important differentiating feature)

Why other options are incorrect:

- Galactosylceramide: Accumulates in Krabbe disease
- Sphingomyelin: Accumulates in Niemann-Pick disease (with hepatosplenomegaly)
- GM1 ganglioside: Accumulates in GM1 gangliosidosis

QUESTION

A 1-year-old child is brought with developmental delay, progressive abdominal distension, and recurrent respiratory infections. On examination, the child has coarse facial features, hepatosplenomegaly, corneal clouding, and skeletal abnormalities. Enzyme assay reveals deficiency of α -L-iduronidase, consistent with Hurler syndrome. Which of the following substances accumulates in this condition?

A Heparan sulfate (HS) and Dermatan sulfate (DS)

CORRECT

B Chondroitin sulfate (CS) and Keratan sulfate (KS)

C Dermatan sulfate (DS) only

D Sphingolipids

RIGHT ANSWER

OK **A. Heparan sulfate (HS) and Dermatan sulfate (DS)**

WHY THIS IS RIGHT

The clinical picture-coarse facial features, organomegaly, corneal clouding, and intellectual disability-with deficiency of α -L-iduronidase is characteristic of Hurler syndrome (Mucopolysaccharidosis type I).

- α -L-iduronidase is required for degradation of glycosaminoglycans (GAGs)
- Deficiency leads to accumulation of:
 - o Heparan sulfate (HS)
 - o Dermatan sulfate (DS)

Mucopolysaccharidoses (MPS):

Disorder Name	Mnemonic	Enzyme Deficiency	GAG(s) Affected	Key Symptoms
Hurler Syndrome Hurler-Scheie Syndrome	Hurler, Scheie, Hurler-Scheie Syndrome	α -L-Iduronidase	Dermatan Sulfate, Heparan Sulfate	Coarse facial features, corneal clouding, hepatosplenomegaly, skeletal abnormalities
Hunter Syndrome	Hunter	Iduronate sulfatase	Dermatan Sulfate, Heparan Sulfate	Mild facial features
Sandhoff Syndrome A	Sandhoff	Heparan sulfate 6-sulfatase (HSP6S)	Heparan Sulfate	Developmental delay, corneal clouding
Sandhoff Syndrome B	Sandhoff	α -N-Acetylglucosaminidase	Heparan Sulfate	Similar to MPS IIA
Sandhoff Syndrome C	Sandhoff	α -Glucuronidase	Heparan Sulfate	Similar to MPS IIA
Sandhoff Syndrome D	Sandhoff	N-Acetylglucosaminase 6-sulfatase	Heparan Sulfate	Similar to MPS IIA
Morquio Syndrome A	Morquio	Glucosaminase 6-sulfatase	Keratan Sulfate, Chondroitin 6-sulfate	Normal lifespan, short stature
Morquio Syndrome B	Morquio	B-Galactosidase	Keratan Sulfate	Similar to MPS IIA
Hurler-Scheie-Langer Syndrome	Hurler-Scheie-Langer	N-Acetylglucosaminase 6-sulfatase (HSP6S)	Dermatan Sulfate, Heparan Sulfate, Chondroitin 6-sulfate	Normal lifespan, short stature, skeletal abnormalities
By Syndrome	By Syndrome	α -Chondroitinase	Dermatan Sulfate, Heparan Sulfate, Chondroitin 6-sulfate	Normal lifespan, short stature, skeletal abnormalities
Hurler-Scheie Syndrome	Hurler-Scheie	Hydrolase	Hydroxyacid	Joint pain, short stature

QUESTION

A patient presents with xanthomas and elevated cholesterol levels. Laboratory investigations show high LDL but normal triglyceride (TG) levels. There is also a family history of premature myocardial infarction (MI)-the patient's father died due to MI. What is the most likely diagnosis?

A Hyperlipoproteinemia III

B Hyperlipoproteinemia I

C **Hyperlipoproteinemia IIa**

CORRECT

D Hyperlipoproteinemia IIb

RIGHT ANSWER

OK **C. Hyperlipoproteinemia IIa**

WHY THIS IS RIGHT

The clinical picture-tendon xanthomas, elevated LDL cholesterol with normal triglycerides, and family history of premature myocardial infarction-is characteristic of Type IIa hyperlipoproteinemia (familial hypercholesterolemia).

- Mutation in LDL receptor (most common) or ApoB-100
- Leads to reduced clearance of LDL from plasma

Lipid profile:

- ↑ LDL cholesterol
- Normal triglycerides

Clinical features:

- Tendon xanthomas (Achilles, extensor tendons)
- Xanthelasma
- Premature atherosclerosis -> early MI

Why other options are incorrect:

- Type III (dysbetalipoproteinemia): ↑ cholesterol + ↑ triglycerides, palmar xanthomas
- Type I: ↑ chylomicrons -> ↑ triglycerides (pancreatitis)
- Type IIb: ↑ LDL + ↑ VLDL -> ↑ cholesterol + ↑ triglycerides

QUESTION

A patient presented with orange coloured tonsils, decreased HDL <5 mg/dl, triglyceride 140 mg/dl. What is the most likely diagnosis?

A Tangier disease

CORRECT

B Abetalipoproteinemia

C Familial hypercholesterolemia

D Familial hyperchylomicronemia

RIGHT ANSWER

OK A. Tangier disease

WHY THIS IS RIGHT

Tangier disease is a rare autosomal recessive disorder caused by a defect in the ABCA1 transporter, which is responsible for efflux of cholesterol from peripheral cells to HDL.

- Impaired cholesterol transport -> failure of HDL formation
- Leads to extremely low HDL levels (<5 mg/dL)
- Cholesterol accumulates in tissues of the reticuloendothelial system

Characteristic clinical features:

- Orange-colored tonsils (pathognomonic clue)
- Very low HDL levels
- Peripheral neuropathy
- Hepatosplenomegaly (sometimes)

Why other options are incorrect:

- Abetalipoproteinemia: Absence of Apo B -> fat malabsorption, acanthocytosis
- Familial hypercholesterolemia: ↑ LDL, tendon xanthomas
- Familial hyperchylomicronemia: ↑ triglycerides, pancreatitis

QUESTION

Which of the following defects causes familial hypercholesterolemia?

A Apo E

B Apo CII

C LDL receptor

CORRECT

D Apo B48

RIGHT ANSWER

OK

C. LDL receptor

WHY THIS IS RIGHT

Familial hypercholesterolemia (FH) is most commonly caused by a defect in the LDL receptor, which is responsible for removing low-density lipoprotein (LDL) particles from the bloodstream. When the LDL receptor is defective or absent, LDL cannot be efficiently taken up by the liver, leading to markedly elevated plasma LDL cholesterol levels. This results in premature atherosclerosis, tendon xanthomas, and an increased risk of early coronary artery disease.

Other options are associated with different lipid disorders:

- Apo E defect -> Type III hyperlipoproteinemia
- Apo CII defect -> Impaired lipoprotein lipase activation (Type I hyperlipoproteinemia)
- Apo B48 -> Involved in chylomicron formation, not FH.

Type	Inherit	Pathogenesis	Blood Level	Clinical
I- Hyper-chylomicronaemia	Ar	Lipoprotein Lipase/ApoC-II deficiency	Chylomicrons, TG, Cholesterol	- Pancreatitis - Eruptive xanthomas - CREAM Supernatant
II- Familial hypercholesterolemia	AD	Absent LDL Receptors/ApoB-100	IIa: LDL IIb: LDL + TG	- Accelerated Atherosclerosis - Tendon Xanthomas - Corneal arcus, Xanthelasma (IIa) - Ruber-eruptive xanthomas (IIb)
III- Dysbeta-lipoproteinemia	Ar	Defective ApoE	Chylomicrons, VLDL, LDL	- Premature atherosclerosis - Tuber-eruptive & Palmar Xanthomas - Broad beta band
IV- Hyper-triglyceridemia	AD	↑↑ Hepatic VLDL	VLDL, TG Type 5: VLDL + chylomicrons	- Broad beta band - Acute pancreatitis - Insulin resistance - Tuber-eruptive Xanthomas

QUESTION

| Statement NOT true about β -oxidation of fatty acids:

- A** CAT I and CAT II transport fatty acids
- B** ATP is used for transport
- C** CoASH released in cytosol during acyl-carnitine formation CORRECT
- D** Carnitine released inside mitochondria

RIGHT ANSWER

- OK** **C. CoASH released in cytosol during acyl-carnitine formation**

WHY THIS IS RIGHT

β -oxidation of fatty acids occurs in the mitochondrial matrix, and long-chain fatty acids require the carnitine shuttle for mitochondrial transport.

Carnitine Shuttle Steps:

1. CPT-I (CAT-I): Converts fatty acyl-CoA $\rightarrow$ acyl-carnitine in the cytosol/outer mitochondrial membrane.
2. Acyl-carnitine is transported into mitochondria.
3. CPT-II (CAT-II): Converts acyl-carnitine $\rightarrow$ fatty acyl-CoA in the mitochondrial matrix, releasing carnitine.

Key points:

- ATP is used earlier for fatty acid activation to fatty acyl-CoA.
- Carnitine is released inside mitochondria after reversion.
- CoA remains within the mitochondrial matrix, not released in the cytosol during acyl-carnitine formation.

QUESTION

| Lorenzo oil is used in the treatment of:

A Fabry's disease

B Adrenoleukodystrophy

CORRECT

C Hurler's disease

D Gaucher disease

RIGHT ANSWER

OK B. Adrenoleukodystrophy

WHY THIS IS RIGHT

Lorenzo's oil is a mixture of oleic acid and erucic acid used to treat X-linked adrenoleukodystrophy (ALD). It works by inhibiting the synthesis of very long-chain fatty acids (VLCFAs), which accumulate in ALD and cause demyelination of the CNS and adrenal insufficiency. Early use can slow disease progression in asymptomatic patients.

Why the others are incorrect:

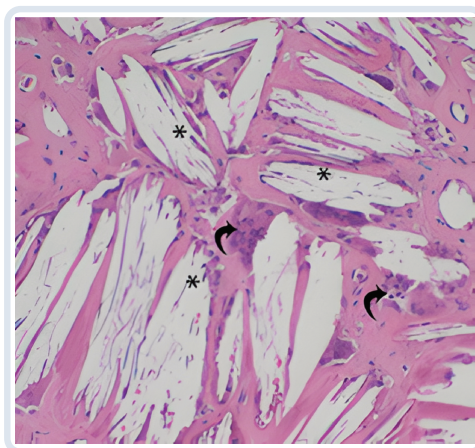
A. Fabry's disease: Treated with enzyme replacement therapy for α -galactosidase A deficiency.

C. Hurler's disease: Managed with enzyme replacement or hematopoietic stem cell transplant for α -L-iduronidase deficiency.

D. Gaucher disease: Treated with glucocerebrosidase enzyme replacement or substrate reduction therapy

QUESTION

A previously healthy 14-year-old boy comes to the physician because of a 5-day history of pain at the back of his left ankle. His mother had a myocardial infarction at 54 years. His BMI is 23 kg/m². A photomicrograph from a biopsy of the nodule is shown. Serum studies in this patient are most likely to show increased levels of which of the following?



A Low-density lipoprotein

CORRECT

B Uric acid

C Triglycerides

D Rheumatoid factor

RIGHT ANSWER

OK **A. Low-density lipoprotein**

WHY THIS IS RIGHT

This patient likely has familial hypercholesterolemia (FH), suggested by tendon xanthomas at the Achilles, strong family history of premature myocardial infarction, and normal BMI. FH causes elevated LDL cholesterol, which deposits in tendons and skin as xanthomas, visible on biopsy. These nodules are cholesterol-rich, not urate crystals.

Why the others are incorrect:

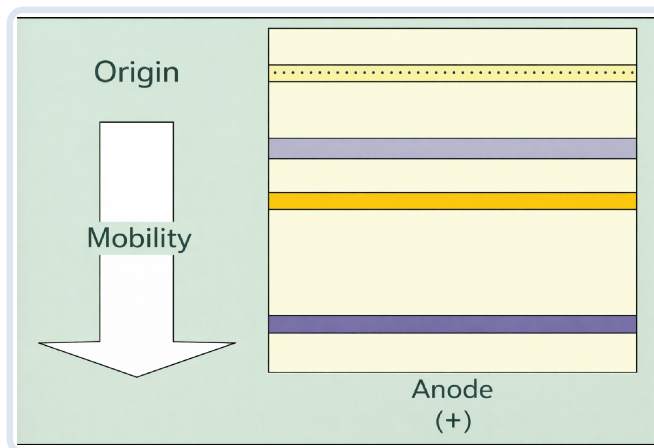
B. Uric acid: Elevated in gout, but tophi in a 14-year-old without acute joint pain is unlikely; biopsy shows lipid, not urate crystals.

C. Triglycerides: Elevated triglycerides cause eruptive xanthomas, which are small papules, not tendon nodules.

D. Rheumatoid factor: Marker for rheumatoid arthritis, which presents with joint inflammation, not tendon xanthomas.

QUESTION

In an electrophoretic separation procedure, a technician observes the migration of lipoproteins from the cathode (-) to the anode (+) side. Referring to the provided electrophoresis image, which of the following correctly orders the lipoproteins from cathode to anode?



A CM > LDL > VLDL > HDL

CORRECT

B CM > VLDL > LDL > HDL

C HDL > VLDL > LDL > CM

D HDL > VLDL > LDL > CM

RIGHT ANSWER

OK A. CM > LDL > VLDL > HDL

WHY THIS IS RIGHT

In lipoprotein electrophoresis from cathode (-) to anode (+), migration depends on size, density, and charge:

- Chylomicrons (CM): Largest and least dense, remain near the origin, migrating least.
- LDL: Moderately dense and negatively charged, migrate more than chylomicrons.
- VLDL: Less dense than LDL, slightly slower migration toward anode.
- HDL: Smallest and densest, migrates farthest toward the anode.

Why the others are incorrect:

B. CM > VLDL > LDL > HDL: Misplaces LDL and VLDL migration order.

C. HDL > VLDL > LDL > CM: Incorrect; HDL moves fastest toward anode, not first in sequence from cathode.

QUESTION

A 45-year-old woman undergoes routine health screening, including a lipid profile, while fasting. Her results show a total cholesterol of 300 mg/dL, HDL cholesterol of 25 mg/dL, and triglycerides of 150 mg/dL. Utilizing the Friedewald equation to assess her cardiovascular risk, what is the LDL cholesterol level in this patient?

A 245 mg/dl

CORRECT

B 135 mg/dl

C 85 mg/dl

D 45 mg/dl

RIGHT ANSWER

OK A. 245 mg/dl

WHY THIS IS RIGHT

Use the Friedewald equation (valid when TG < 400 mg/dL and patient is fasting):
 $LDL = TC - HDL - \frac{TG}{5}$

Given values

- Total cholesterol (TC) = 300 mg/dL
- HDL = 25 mg/dL
- Triglycerides (TG) = 150 mg/dL

Step 1: Calculate VLDL

Step 2: Calculate LDL

Correct answer: a) 245 mg/dL

Remember:

- Friedewald formula: $LDL = TC - HDL - (TG/5)$
- TG/5 approximates VLDL cholesterol
- Not valid if TG > 400 mg/dL or non-fasting sample.

$$LDL = \text{Total Cholesterol} - HDL - \frac{\text{Triglycerides}}{5}$$

$$VLDL = \frac{TG}{5} = \frac{150}{5} = 30$$

$$LDL = 300 - 25 - 30$$

$$LDL = 245 \text{ mg/dL}$$

QUESTION

An 18-year-old man comes to the office due to a progressive skin rash over the past year. He also has a long-standing history of an intermittent burning sensation in his palms and soles that is exacerbated by stress and fatigue. The burning sensation is particularly severe after exercise, during which the patient notes that he sweats minimally. Laboratory evaluation reveals an undetectable level of α -galactosidase A. Which of the following conditions is this patient at greatest risk for developing?

A Ataxia

B Hepatomegaly

C Optic atrophy

D Renal failure

CORRECT

RIGHT ANSWER

OK D. Renal failure

WHY THIS IS RIGHT

This patient has Fabry disease, an X-linked lysosomal storage disorder caused by α -galactosidase A deficiency, leading to accumulation of globotriaosylceramide in various tissues. Early symptoms include angiokeratomas, acroparesthesias (burning in hands/feet), and hypohidrosis. Over time, deposition in renal vasculature and glomeruli causes progressive renal insufficiency and eventual renal failure if untreated.

Why the others are incorrect:

A. Ataxia: Not a primary feature of Fabry disease; neurologic involvement is mainly neuropathic pain.

B. Hepatomegaly: Liver enlargement is uncommon in Fabry disease.

C. Optic atrophy: Ocular manifestations usually involve cornea verticillata, not optic nerve degeneration.

QUESTION

A 56-year-old man suffering from uncontrolled diabetes mellitus came for a routine checkup. His lipid profile revealed increased TG (triacylglycerol) and VLDL (very low-density lipoprotein). What is the cause of this abnormal lipid profile?

A Increased activity of lipoprotein lipase and decreased activity of hormone sensitive lipase

B Increased activity of hormone sensitive lipase and decreased lipoprotein lipase activity

CORRECT

C Increase in peripheral function of LDL receptors

D Increased in activity of hepatic lipase

RIGHT ANSWER

OK **B. Increased activity of hormone sensitive lipase and decreased lipoprotein lipase activity**

WHY THIS IS RIGHT

- Hormone-sensitive lipase (HSL): Breaks down stored triglycerides in adipose tissue into free fatty acids, which are transported to the liver. Increased HSL activity leads to more fatty acids reaching the liver, increasing VLDL synthesis.
- Lipoprotein lipase (LPL): Hydrolyzes circulating triglycerides in VLDL and chylomicrons. Decreased LPL activity impairs VLDL clearance, resulting in hypertriglyceridemia.

Why other options are incorrect:

- A. Increased LPL & decreased HSL: Would lower triglycerides and VLDL, opposite effect.
- C. Increase in LDL receptor function: Lowers LDL cholesterol, not VLDL or TG.
- D. Increased hepatic lipase: Mainly affects HDL and IDL metabolism, not primary VLDL/TG elevation.

QUESTION

A 10-year-old boy with persistent fatigue presents with fatigue and biopsy of muscle shows accumulation of triglycerides esterified with long chain fatty acid (LCFA). Which of the following is the diagnosis?

A Carnitine deficiency

CORRECT

B FA synthase defect

C LPL defect

D LDL receptor defect

RIGHT ANSWER

OK **A. Carnitine deficiency**

WHY THIS IS RIGHT

The patient shows muscle triglyceride accumulation with long-chain fatty acids (LCFA), suggesting an impairment in mitochondrial fatty acid transport or oxidation. Carnitine deficiency prevents the transport of LCFA into mitochondria for β -oxidation, leading to lipid accumulation in muscle, exercise intolerance, and fatigue.

Why other options are incorrect:

B. FA synthase defect: Would reduce fatty acid synthesis, not cause accumulation.

C. Lipoprotein lipase (LPL) defect: Causes hypertriglyceridemia in plasma, not muscle lipid deposition.

D. LDL receptor defect: Causes elevated plasma LDL, not intracellular muscle lipid accumulation.

QUESTION

Which of the following causes the inactivation of acetyl-CoA carboxylase?

A Phosphorylation

CORRECT

B Insulin

C High levels of citrate

D Dephosphorylation

RIGHT ANSWER

OK **A. Phosphorylation**

WHY THIS IS RIGHT

Acetyl-CoA carboxylase (ACC) is the rate-limiting enzyme of fatty acid synthesis, converting acetyl-CoA → malonyl-CoA. Phosphorylation by AMP-activated protein kinase (AMPK) inactivates ACC, decreasing fatty acid synthesis during low-energy states.

Why other options are incorrect:

- B. Insulin: Activates ACC by stimulating dephosphorylation, promoting fatty acid synthesis.
- C. High levels of citrate: Allosterically activate ACC, counteracting phosphorylation.
- D. Dephosphorylation: Activates ACC, not inactivates it.

QUESTION

A 12-year-old girl suffers from an inadequately treated type 1 diabetes mellitus. She is currently hyperglycemic. Which of the following is most likely to occur?

- A** Glycogenesis in muscle
- B** Increased protein synthesis
- C** Increased conversion of fatty acids to acetyl-CoA
- D** Decreased cholesterol synthesis

CORRECT

RIGHT ANSWER

- OK** **C. Increased conversion of fatty acids to acetyl-CoA**

WHY THIS IS RIGHT

In poorly controlled type 1 diabetes mellitus, insulin deficiency causes a catabolic state. Low insulin and high glucagon activate hormone-sensitive lipase, increasing lipolysis. Released fatty acids are transported to the liver and undergo β -oxidation, producing large amounts of acetyl-CoA, which contributes to ketone body formation during hyperglycemia.

Why other options are incorrect:

- A. Glycogenesis in muscle: Requires insulin; it is decreased in insulin deficiency.
- B. Increased protein synthesis: Insulin is anabolic; protein breakdown predominates instead.
- D. Decreased cholesterol synthesis: Cholesterol synthesis is actually reduced due to low insulin, but the dominant metabolic change here is increased fatty acid oxidation

QUESTION

A 8-year-old boy is brought to the emergency department due to vomiting and lethargy. He hasn't eaten anything for 24 hours. On examination, the patient appears listless. Mild hepatomegaly is noted.

Laboratory results are as follows:

Glucose- 22mg/dl

Acetoacetate - Undetected

ALT-47 IU

AST-50 IU

Patient began seizing in the emergency. The likely deficient enzyme is ?

A Acetyl-CoA carboxylase

B Glycogen phosphorylase

C Acyl-CoA dehydrogenase

CORRECT

D Glucose 6-phosphatase

RIGHT ANSWER

OK C. Acyl-CoA dehydrogenase

WHY THIS IS RIGHT

This child has severe hypoglycemia with absent ketone bodies after fasting, along with vomiting, lethargy, seizures, and mild hepatomegaly-classic for fatty acid oxidation defect, most commonly medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. In fasting states, inability to perform β -oxidation leads to failure of ketogenesis and inadequate energy production, causing hypoketotic hypoglycemia and neurologic symptoms.

Why other options are incorrect:

A. Acetyl-CoA carboxylase: Involved in fatty acid synthesis, not fasting metabolism.

B. Glycogen phosphorylase: Deficiency causes ketotic hypoglycemia with normal FA oxidation.

D. Glucose-6-phosphatase: Von Gierke disease shows lactic acidosis and ketosis, not absent ketones.

QUESTION

| The enzyme that is not used in cholesterol synthesis?

- A** Thiolase
- B** HMG CoA reductase
- C** HMG CoA synthase
- D** **HMG CoA lyase**

CORRECT

RIGHT ANSWER

- OK** **D. HMG CoA lyase**

WHY THIS IS RIGHT

Cholesterol synthesis occurs in the cytosol and ER and proceeds via formation of HMG-CoA, which is then reduced to mevalonate. HMG-CoA lyase is not involved in cholesterol synthesis; instead, it functions in ketone body synthesis, where it cleaves HMG-CoA to acetoacetate and acetyl-CoA in liver mitochondria.

Why other options are incorrect:

- A. Thiolase: Catalyzes condensation of acetyl-CoA molecules in early cholesterol synthesis.
- B. HMG-CoA reductase: Rate-limiting enzyme converting HMG-CoA to mevalonate.
- C. HMG-CoA synthase: Required to form HMG-CoA from acetyl-CoA and acetoacetyl-CoA.

QUESTION

A 30-year-old man is being evaluated for a deficiency of lipoprotein lipase and on further investigation, he too was diagnosed with the same condition. Which of the following lipoproteins will be elevated in this patient's plasma?

A Chylomicron and Triglycerides

CORRECT

B LDL and HDL

C HDL and VLDL

D Chylomicron and LDL

RIGHT ANSWER

OK

A. Chylomicron and Triglycerides

WHY THIS IS RIGHT

Lipoprotein lipase (LPL) hydrolyzes triglycerides in chylomicrons and VLDL into free fatty acids for uptake by tissues. In LPL deficiency, this step is impaired, leading to accumulation of chylomicrons in plasma and marked hypertriglyceridemia. Patients classically present with milky plasma, pancreatitis, eruptive xanthomas, and lipemia retinalis, while cholesterol-rich lipoproteins are less affected.

Why other options are incorrect:

B. LDL and HDL: These are not substrates for LPL.

C. HDL and VLDL: VLDL rises, but HDL typically decreases.

D. Chylomicron and LDL: LDL metabolism is not directly dependent on LPL.

QUESTION

Which clinical feature helps distinguish Refsum disease from Zellweger syndrome?

- A** Hearing loss
- B** Retinitis pigmentosa
- C** Neonatal hypotonia
- D** Craniofacial dysmorphism

CORRECT

RIGHT ANSWER

- OK** **B. Retinitis pigmentosa**

WHY THIS IS RIGHT

Refsum disease classically presents with retinitis pigmentosa, peripheral neuropathy, cerebellar ataxia, and hearing loss due to phytanic acid accumulation, while neonatal presentation is absent. In contrast, Zellweger syndrome presents in the neonatal period with hypotonia, seizures, craniofacial dysmorphism, and early death, and retinal changes are not a defining early feature. Thus, retinitis pigmentosa helps distinguish Refsum disease from Zellweger syndrome.

Why other options are incorrect:

- A. Hearing loss: Seen in Refsum disease but not exclusive.
- C. Neonatal hypotonia: Typical of Zellweger, not Refsum.
- D. Craniofacial dysmorphism: Characteristic of Zellweger syndrome.

QUESTION

A 10-year-old boy with knock knees and a waddling gait has severe skeletal deformities, mild hepatomegaly, and corneal clouding. Unlike most mucopolysaccharidoses, his intellect is completely normal. Which MPS subtype is most likely?

A Hurler syndrome

B Hunter syndrome

C Sanfilippo syndrome

D Morquio syndrome

CORRECT

RIGHT ANSWER

OK **D. Morquio syndrome**

WHY THIS IS RIGHT

Morquio syndrome (MPS IV) is characterized by severe skeletal dysplasia (knock knees, short stature, waddling gait), corneal clouding, and sometimes mild hepatomegaly, with normal intelligence, which is its key distinguishing feature among MPS disorders. It results from defective degradation of keratan sulfate, leading predominantly to bone involvement rather than CNS damage.

Why other options are incorrect:

A. Hurler syndrome: Causes severe intellectual disability, coarse facies, and early death.

B. Hunter syndrome: No corneal clouding and usually has cognitive impairment.

C. Sanfilippo syndrome: Predominantly neurodegenerative with severe intellectual decline, minimal skeletal changes.

QUESTION

A 17-year-old tall male presents for a routine check-up. He has long limbs, arachnodactyly, hypermobile joints, a high-arched palate, and an increased arm span. Genetic testing reveals a defect in the FBN1 gene. Which of the following is the most likely diagnosis?

A Ehlers-Danlos Syndrome

B Marfan Syndrome

CORRECT

C Osteogenesis Imperfecta

D Heritable Disorders of Connective Tissue

RIGHT ANSWER

OK B. Marfan Syndrome

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$19” here and does not include a written explanation for this item.

QUESTION

| Respiratory distress syndrome is caused by the deficiency of?

- A** Cephalin
- B** Sphingomyelin
- C** **Lecithin**
- D** Cardiolipin

CORRECT

RIGHT ANSWER

OK **C. Lecithin**

WHY THIS IS RIGHT

Respiratory Distress Syndrome (RDS) in newborns is caused by deficiency of pulmonary surfactant, the most important component of which is lecithin (dipalmitoyl phosphatidylcholine, DPPC).

Function of lecithin (surfactant):

- Reduces alveolar surface tension
- Prevents alveolar collapse (atelectasis) during expiration
- Maintains lung compliance

Clinical correlation:

- Common in premature infants (immature type II pneumocytes)
- Assessed by Lecithin : Sphingomyelin (L/S) ratio in amniotic fluid

o L/S ratio < 2 -> high risk of RDS

Why other options are incorrect:

- Cephalin: Phospholipid involved in coagulation
- Sphingomyelin: Used as a reference in L/S ratio, not the main surfactant
- Cardiolipin: Found in mitochondrial membranes

QUESTION

An infant is brought by his parents with complaints that his urine turns black on standing. Which of the following metabolic disorders is likely?

- A Phenylketonuria
- B Alkaptonuria**
- C Homocystinuria
- D Maple syrup urine disease

CORRECT

RIGHT ANSWER

- B. Alkaptonuria**

WHY THIS IS RIGHT

Alkaptonuria is an autosomal recessive disorder of tyrosine metabolism caused by deficiency of the enzyme homogentisate oxidase.

Clinical features:

- Dark/black urine on standing (early clue in infants)
- Ochronosis (bluish-black pigmentation of connective tissue)
- Arthritis in adulthood

Why other options are incorrect:

- Phenylketonuria (PKU): Musty/mousy odor urine, intellectual disability
- Homocystinuria: Marfanoid habitus, lens dislocation (downward)
- Maple syrup urine disease: Sweet “maple syrup” odor of urine

QUESTION

An adult patient presents with tall stature, pigeon chest deformity, long arm span, lens dislocation, and cardiac defects. This patient is likely to have a defect in which of the following?

A Elastase

B Fibrillin

CORRECT

C Collagen

D Laminin

RIGHT ANSWER

OK

B. Fibrillin

WHY THIS IS RIGHT

The clinical features-tall stature, long arm span, pectus deformity (pigeon chest), lens dislocation, and cardiac abnormalities (aortic root dilation)-are classic for Marfan syndrome.

- Mutation in fibrillin-1 (FBN1 gene)
- Fibrillin is a key component of microfibrils in connective tissue

Clinical features explained:

- Skeletal: Tall, long limbs (arachnodactyly), pectus deformity
- Ocular: Lens dislocation (typically upward)
- Cardiac: Aortic root dilation -> risk of aortic aneurysm/dissection

Why other options are incorrect:

- Elastase: Enzyme, not structural protein defect
- Collagen: Seen in Ehlers-Danlos or osteogenesis imperfecta
- Laminin: Basement membrane component

QUESTION

A 26-year-old male presented with colicky pain. Following type of crystals were found on urine examination. What is the likely diagnosis?



- A** ADPKD
- B** Calcium oxalate stones
- C** Cystinuria
- D** Glomerulonephritis

CORRECT

RIGHT ANSWER

OK C. Cystinuria

WHY THIS IS RIGHT

The presence of hexagonal crystals in urine is pathognomonic of cystinuria, a genetic disorder affecting amino acid transport. Defective renal tubular reabsorption of cystine and dibasic amino acids (COLA: cystine, ornithine, lysine, arginine)

- Cystine is poorly soluble in urine -> precipitates to form hexagonal crystals and stones
- Leads to recurrent renal colic and kidney stones

Clinical features:

- Recurrent kidney stones at a young age
- Colicky abdominal pain
- May have family history (autosomal recessive)

Why other options are incorrect:

- Calcium oxalate stones: Envelope-shaped crystals
- ADPKD: Cystic kidneys, not crystal-related
- Glomerulonephritis: Hematuria, proteinuria (no characteristic crystals)

QUESTION

Which of the following is deficient in a patient with phenylketonuria with normal phenylalanine hydroxylase levels?

A Dihydrobiopterin reductase

CORRECT

B Tyrosine transaminase

C Dopa decarboxylase

D Tyrosine hydroxylase

RIGHT ANSWER

OK A. Dihydrobiopterin reductase

WHY THIS IS RIGHT

In a patient with hyperphenylalaninemia but normal phenylalanine hydroxylase (PAH), the defect lies in the cofactor tetrahydrobiopterin (BH₄) metabolism.

Key enzyme involved: Dihydrobiopterin reductase (DHPR) Deficiency -> ↓ BH₄ availability

- Impaired phenylalanine -> tyrosine conversion -> ↑ phenylalanine
- Also affects neurotransmitter synthesis:

- o ↓ dopamine (tyrosine pathway)
- o ↓ serotonin (tryptophan pathway)

Clinical importance:

- More severe than classical PKU
- Presents with neurological deficits due to neurotransmitter deficiency

Why other options are incorrect:

- Tyrosine transaminase: Involved in tyrosine metabolism (tyrosinemia)
- Dopa decarboxylase: Involved in dopamine synthesis, not primary cause here
- Tyrosine hydroxylase: Requires BH₄ but is not the defective enzyme in this condition

QUESTION

A patient has markedly elevated plasma phenylalanine levels and low tyrosine levels. Phenylalanine hydroxylase enzyme assay is normal. Which of the following cofactor deficiencies is most likely responsible for this presentation?

A Biopterin

CORRECT

B DHFR

C PLP

D THF

RIGHT ANSWER

OK A. Biopterin

WHY THIS IS RIGHT

Conversion of phenylalanine → tyrosine requires:

- Enzyme: Phenylalanine hydroxylase (PAH)
- Cofactor: Tetrahydrobiopterin (BH₄, biopterin)

In this case, PAH is normal, so the defect lies in the cofactor (BH₄) deficiency, leading to:

- ↑ Phenylalanine
- ↓ Tyrosine

BH₄ is also required for synthesis of neurotransmitters:

- Dopamine (via tyrosine hydroxylase)
- Serotonin (via tryptophan hydroxylase)

Thus, deficiency leads to: Neurological symptoms more severe than classical PKU

Why other options are incorrect:

- DHFR: Involved in folate metabolism
- PLP (Vitamin B6): Cofactor for transamination reactions
- THF: Involved in one-carbon metabolism (DNA synthesis)

QUESTION

2, 3 BPG binds to:

- A** Haem part of Hb
- B** Globin part of Hb CORRECT
- C** Haem part of Myoglobin
- D** Globin part of Myoglobin

RIGHT ANSWER

- OK** **B. Globin part of Hb**

WHY THIS IS RIGHT

2,3-Bisphosphoglycerate (2,3-BPG) binds to the positively charged pocket formed by the β -globin chains of deoxyhemoglobin, not to the heme group. This interaction stabilizes the T (tense) state of hemoglobin, thereby reducing hemoglobin's affinity for oxygen and promoting oxygen release to peripheral tissues.

Why other options are incorrect:

- Heme part of Hb: Oxygen binds to the iron in the heme group; 2,3-BPG does not bind here.
- Heme part of Myoglobin: Myoglobin has a single heme and does not interact with 2,3-BPG.
- Globin part of Myoglobin: Myoglobin lacks the central cavity present in hemoglobin and therefore cannot bind 2,3-BPG.

QUESTION

| Which diagnostic tool is preferred for metabolic disease screening in children?

A Tandem mass spectrometry

CORRECT

B Complete blood count (CBC)

C Urinalysis

D Genetic testing

RIGHT ANSWER

OK **A. Tandem mass spectrometry**

WHY THIS IS RIGHT

Tandem mass spectrometry (MS/MS) is the preferred screening tool for inborn errors of metabolism in newborns and children. It can simultaneously detect multiple metabolic disorders by measuring amino acids, organic acids, and acylcarnitines from a single dried blood spot.

This method is widely used in newborn screening programs to identify disorders such as phenylketonuria, maple syrup urine disease, fatty acid oxidation defects, and organic acidemias, allowing early diagnosis and treatment to prevent severe complications.

Why not other options:

- CBC: Evaluates blood cells (RBCs, WBCs, platelets); not useful for detecting metabolic enzyme defects.
- Urinalysis: May detect some metabolites but is not sensitive or comprehensive for screening multiple metabolic disorders.
- Genetic testing: Confirms specific mutations but is not practical for large-scale initial screening of many metabolic diseases.

QUESTION

Which of the following is not an amino acid-derived neurotransmitter?

A Dopamine

B Serotonin

C GABA

D **Creatine**

CORRECT

RIGHT ANSWER

OK **D. Creatine**

WHY THIS IS RIGHT

Many neurotransmitters are derived from amino acids or their metabolites.

- Dopamine is synthesized from the amino acid tyrosine.
- Serotonin (5-HT) is derived from tryptophan.
- GABA (γ -aminobutyric acid) is produced from glutamate by the enzyme glutamate decarboxylase.

Creatine, however, is not a neurotransmitter. It is a high-energy storage molecule synthesized from glycine, arginine, and methionine and is mainly involved in energy metabolism in muscle and brain.

QUESTION

Which is not a product of heme catabolism?

A Ferrous ion

B Aminolaevulinic acid

CORRECT

C Biliverdin

D Carbon monoxide

RIGHT ANSWER

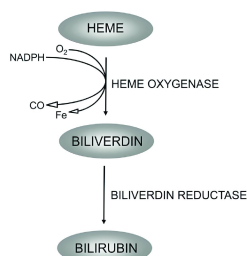
OK B. Aminolaevulinic acid

WHY THIS IS RIGHT

During heme catabolism, heme is degraded by the enzyme heme oxygenase to produce biliverdin, ferrous iron (Fe^{2+}), and carbon monoxide (CO). Biliverdin is subsequently reduced to bilirubin by biliverdin reductase.

Aminolevulinic acid (ALA) is not a product of heme breakdown; instead, it is an intermediate in heme synthesis, formed from glycine and succinyl-CoA by ALA synthase in mitochondria.

Therefore, aminolevulinic acid belongs to the heme biosynthesis pathway, not the catabolic pathway, making it the correct answer.



QUESTION

Match the following Amino acid products:

A. Tyrosine	1. Melatonin
B. Tryptophan	2. Glutathione
C. Glycine	3. Nor epinephrine
D. Histidine	4. Histamine

A A-2, B-4, C-3, D-1

B A-3, B-1, C -2, D-4

CORRECT

C A-1, B-2, C-3, D-4

D A-4, B-3, C-2, D-1

RIGHT ANSWER

OK B. A-3, B-1, C -2, D-4

WHY THIS IS RIGHT

Amino Acid Conversions

Tyrosine (A)	Norepinephrine (3)
Tryptophan (B)	Melatonin (1)
Glycine (C)	Glutathione (2)
Histidine (D)	Histamine (4)

QUESTION

Sodium benzoate is given for urea cycle defects for scavenging ammonia. Which amino acid donates amino group to the scavengers?

A Arginine

B Glycine

CORRECT

C Glutamine

D Aspartate

RIGHT ANSWER

B. Glycine

WHY THIS IS RIGHT

In urea cycle disorders, toxic ammonia accumulates because the body cannot efficiently convert ammonia to urea. Nitrogen scavenger drugs such as sodium benzoate are used to remove excess nitrogen through alternative metabolic pathways.

Mechanism of Sodium Benzoate:

- Sodium benzoate conjugates with glycine in the liver.
- This reaction forms hippuric acid (benzoylglycine).
- Hippuric acid is then excreted in urine, removing one nitrogen atom from the body.

Drug	Amino acid used	Product excreted
Sodium benzoate	Glycine	Hippurate
Sodium phenylacetate / phenylbutyrate	Glutamine	Phenylacetylglutamine

Sodium benzoate | Glycine | Hippurate |

Sodium phenylacetate / phenylbutyrate | Glutamine | Phenylacetylglutamine |

QUESTION

Match the following amino acids with their respective derivatives:

1. Tyrosine
2. Tryptophan
3. Glycine
4. Cysteine
- a. Thyroxine
- b. Taurine
- c. Creatine
- d. Melatonin

A 1-a, 2-d, 3-c, 4-b

CORRECT

B 1-d, 2-a, 3-b, 4-c

C 1-c, 2-b, 3-d, 4-a

D 1-b, 2-c, 3-a, 4-d

RIGHT ANSWER

OK A. 1-a, 2-d, 3-c, 4-b

WHY THIS IS RIGHT

Amino Acid	Important Derivative	Key Function
Tyrosine	Thyroxine (T4)	Thyroid hormone involved in regulation of metabolism
Tryptophan	Melatonin	Hormone regulating circadian rhythm and sleep
Glycine	Creatine	Involved in energy storage in muscles (phosphocreatine system)
Cysteine	Taurine	Important component of bile salts and antioxidant functions

- Tyrosine -> Thyroid hormones (T3, T4), catecholamines, melanin
- Tryptophan -> Serotonin -> Melatonin
- Glycine + Arginine + Methionine -> Creatine
- Cysteine -> Taurine -> Bile salts

QUESTION

A 1-day-old boy is diagnosed with hyperphenylalaninemia by newborn screening. He is placed on a special phenylalanine-restricted diet with tyrosine supplementation. The parents are extensively counseled on the boy's condition and informed of the necessary dietary restrictions. They are also instructed to return to his physician for regular follow-up visits. Several months later, laboratory test results indicate that the infant has a normal serum phenylalanine level. Careful examination, however, reveals some neurological abnormalities, including axial hypotonia and microcephaly. Further workup is notable for elevated prolactin, and his physician suspects a cofactor deficiency. Which of the following enzymes is most likely deficient in this patient?

- A** Dihydrobiopterin reductase
- B** Dopamine β -hydroxylase
- C** Phenylalanine hydroxylase
- D** Phenylethanolamine N-methyltransferase

CORRECT

RIGHT ANSWER

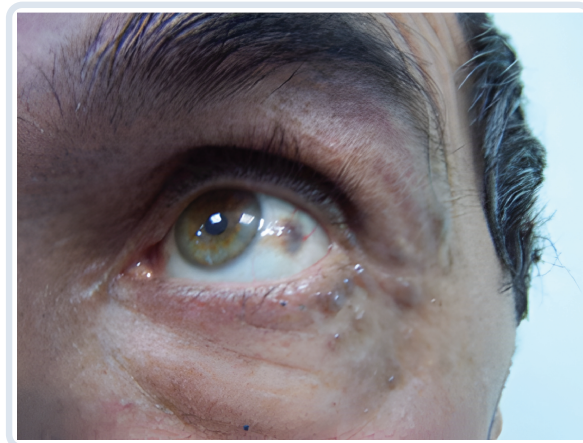
OK A. Dihydrobiopterin reductase

WHY THIS IS RIGHT

The source quiz contains the placeholder "\$1a" here and does not include a written explanation for this item.

QUESTION

A 48-year-old man came with complaints of long-standing arthralgia in the knee. On examination, bluish-black scleral patches were seen as shown. On further inquiry, he says that the urine turns black on exposure to air. Accumulation of which of the following compounds caused this condition?



A Benzoquinone acetate

CORRECT

B Methyl malonyl CoA

C Orotic acid

D Ceramide

RIGHT ANSWER

OK A. Benzoquinone acetate

WHY THIS IS RIGHT

This patient has alkaptonuria, an autosomal recessive disorder caused by homogentisate 1,2-dioxygenase deficiency, leading to accumulation of homogentisic acid. On oxidation, homogentisic acid is converted to benzoquinone acetate, which deposits in connective tissues (ochronosis, bluish-black sclera) and causes urine to darken on standing. Long-term accumulation leads to arthropathy.

Why the others are incorrect:

B. Methylmalonyl CoA: Accumulates in methylmalonic acidemia, causing metabolic acidosis and developmental delay, not dark urine or scleral pigmentation.

C. Orotic acid: Accumulates in orotic aciduria, leading to megaloblastic anemia, not ochronosis.

D. Ceramide: Accumulates in sphingolipidoses (e.g., Farber disease), causing neurologic and skin manifestations, not black urine.

QUESTION

A 7-year-old child was evaluated for night blindness. Ophthalmoscopy showed atrophic changes in the choroid, retina, and pigment epithelium. Urine and plasma ornithine levels were elevated. Defective catabolism of which of the following amino acids led to this condition?

A Histidine

B Glycine

C Serine

D Arginine

CORRECT

RIGHT ANSWER

OK D. Arginine

WHY THIS IS RIGHT

This child has hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome or more specifically juvenile gyrate atrophy of the choroid and retina, suggested by night blindness, retinal/choroidal atrophy, and elevated plasma/urine ornithine. The condition is caused by defective ornithine aminotransferase, which is involved in the catabolism of arginine → ornithine → downstream metabolites. Accumulation of ornithine leads to retinal toxicity and progressive vision loss.

Why the others are incorrect:

A. Histidine: Defects cause urocanic aciduria or histidinemia, not retinal degeneration.

B. Glycine: Defects cause nonketotic hyperglycinemia, leading to neurologic symptoms, not night blindness.

C. Serine: Defects cause serine biosynthesis disorders with neurologic manifestations, not retinal atrophy.

QUESTION

A 3-year-old boy is brought to the physician by his parents because he has not yet begun to walk or speak. Assessment of his developmental milestones shows severe intellectual disability. He dies 6 months later from refractory seizures resulting in respiratory failure. Autopsy shows pallor of the substantia nigra, locus ceruleus, and vagal nucleus dorsalis. The underlying condition responsible for this patient's death is most likely caused by a deficiency of which of the following enzymes?

A Branched chain ketoacid dehydrogenase

B Dopamine hydroxylase

C Homogentisic acid oxidase

D Phenylalanine hydroxylase

CORRECT

RIGHT ANSWER

OK D. Phenylalanine hydroxylase

WHY THIS IS RIGHT

This child's presentation is classic for untreated phenylketonuria (PKU): severe intellectual disability, developmental delay, seizures, and early death if untreated. PKU is caused by a deficiency of phenylalanine hydroxylase, which converts phenylalanine → tyrosine. Accumulation of phenylalanine and its metabolites leads to neurotoxicity, affecting brain development and function, including catecholaminergic regions like the substantia nigra and locus ceruleus, as seen on autopsy.

Why the others are incorrect:

A. Branched-chain ketoacid dehydrogenase: Causes maple syrup urine disease with metabolic acidosis and odor, not this neurologic pattern.

B. Dopamine hydroxylase: Deficiency causes autonomic dysfunction, hypotension, and low norepinephrine, but not severe intellectual disability from birth.

C. Homogentisic acid oxidase: Causes alkaptonuria with dark urine and connective tissue pigmentation; neurologic deficits are not typical.

QUESTION

A group of investigators are studying the regulation of catecholamine synthesis in response to severe stress. In the experiments, subject rats are randomly assigned to either an experimental or a control group. The experimental rats undergo resection of the pituitary gland, and the control rats undergo craniotomy without pituitary resection. The experimental animals are subsequently found to have decreased production of epinephrine by the adrenal medulla and cortisol from the adrenal cortex compared with the control animals. Decrease d activity of which of the following enzymes is most likely responsible for the lower epinephrine in the experimental animals?

A Catechol-O-methyl transferase

B Phenyl ethanolamine-N-methyltransferase

CORRECT

C Dopamine beta-hydroxylase

D Monoamine oxidase

RIGHT ANSWER

OK **B. Phenyl ethanolamine-N-methyltransferase**

WHY THIS IS RIGHT

The pituitary gland produces ACTH, which stimulates the adrenal cortex to release cortisol. Cortisol, in turn, enters the adrenal medulla and induces PNMT, the enzyme that converts norepinephrine to epinephrine. Pituitary resection reduces ACTH -> lower cortisol -> decreased PNMT activity -> reduced epinephrine synthesis.

Why the others are incorrect:

A. Catechol-O-methyltransferase (COMT): Inactivates catecholamines by methylation; not involved in epinephrine synthesis.

C. Dopamine β -hydroxylase: Converts dopamine to norepinephrine; norepinephrine can still be made, but conversion to epinephrine is impaired in this experiment.

D. Monoamine oxidase (MAO): Degrades catecholamines; its activity does not regulate synthesis.

QUESTION

| Serotonin production involves:

- A** Carboxylation -> Hydroxylation of tryptophan
- B** Hydroxylation -> Carboxylation of tryptophan
- C** **Hydroxylation -> Decarboxylation of tryptophan**
- D** Decarboxylation -> Hydroxylation of tryptophan

CORRECT

RIGHT ANSWER

- OK** **C. Hydroxylation -> Decarboxylation of tryptophan**

WHY THIS IS RIGHT

Serotonin (5-hydroxytryptamine, 5-HT) is synthesized from tryptophan in two main steps:

1. Hydroxylation: Tryptophan is converted to 5-hydroxytryptophan by tryptophan hydroxylase (rate-limiting step; requires tetrahydrobiopterin as a cofactor).
2. Decarboxylation: 5-hydroxytryptophan is then decarboxylated by aromatic L-amino acid decarboxylase to form serotonin.

Why other options are incorrect:

A. Carboxylation -> Hydroxylation / B. Hydroxylation -> Carboxylation / D. Decarboxylation -> Hydroxylation: These sequences are biochemically incorrect; serotonin biosynthesis does not involve carboxylation of tryptophan.

QUESTION

A 25-year-old patient presented with dark urine, which further blackens on exposure to air. What's the enzyme defect in this case?

A Fumaryl acetoacetate hydrolase

B Di Hydroxy Phenyl Acetate Dioxygenase

CORRECT

C Homogentisate Dehydrogenase

D Phenyl alanine hydroxylase

RIGHT ANSWER

OK **B. Di Hydroxy Phenyl Acetate Dioxygenase**

WHY THIS IS RIGHT

The patient's dark urine that blackens on standing is a classic sign of alkaptonuria, which results from a block in the tyrosine degradation pathway. The enzyme homogentisate dioxygenase (also called dihydroxyphenylacetate dioxygenase in some texts) is deficient. This leads to accumulation of homogentisic acid, which is excreted in urine and oxidizes to a dark pigment on exposure to air.

Why other options are incorrect:

- A. Fumarylacetoacetate hydrolase: Deficiency causes tyrosinemia type I with liver and kidney failure, not darkening urine.
- C. Homogentisate dehydrogenase: Sometimes synonymous with B, but in this context, the specific enzyme cited is dihydroxyphenylacetate dioxygenase.
- D. Phenylalanine hydroxylase: Deficiency causes phenylketonuria, not urine darkening.

QUESTION

As part of an experiment, healthy volunteers undergo a 12-hour fast and then drink a solution containing radiolabeled alanine. Consecutive blood samples are drawn every 15 minutes for the next 3 hours. Initial blood samples detect the radiolabeled alanine, but analysis of later samples shows that the radiotracer is present in blood primarily in the form of glucose. Before alanine can be converted to glucose, its amino group is transferred to which of the following?

A α -Ketoglutarate

CORRECT

B L-citrulline

C Malate

D Citrate

RIGHT ANSWER

OK A. α -Ketoglutarate

WHY THIS IS RIGHT

Alanine undergoes transamination in the liver as the first step of gluconeogenesis. Its amino group is transferred to α -ketoglutarate, forming glutamate and leaving behind pyruvate, which can then enter gluconeogenesis to produce glucose (as observed in the radiolabel experiment). This process is catalyzed by alanine aminotransferase (ALT).

Why other options are incorrect:

L-citrulline: Intermediate in the urea cycle; not directly involved in alanine transamination.

Malate & Citrate: Intermediates in the TCA cycle, not amino group acceptors for alanine.

QUESTION

A 24-month-old boy is brought to the physician for the evaluation of intellectual and behavioral abnormalities and abnormal movements of his extremities. His mother reports that he often hits his head and limbs against furniture. He is unable to walk without support and speaks in unclear 2-word phrases. Examination shows multiple bruises on the forehead and several healing wounds over the fingers. There is increased muscle tone in all extremities. Laboratory studies show an increased serum uric acid concentration. The serum concentration of which of the following substances is most likely to also be increased in this patient?

A HGPRT

B Phosphoribosyl pyrophosphate

CORRECT

C Deoxyadenosine triphosphate

D Branched-chain amino acids

RIGHT ANSWER

OK

B. Phosphoribosyl pyrophosphate

WHY THIS IS RIGHT

This child has Lesch-Nyhan syndrome, caused by deficiency of hypoxanthine-guanine phosphoribosyltransferase (HGPRT). HGPRT normally salvages hypoxanthine and guanine to IMP and GMP. Its deficiency leads to:

- Hyperuricemia (excess purine degradation)
- Self-mutilation, intellectual disability, dystonia, spasticity

The lack of purine salvage causes PRPP accumulation, because it is not being used in the salvage pathway. Elevated PRPP stimulates de novo purine synthesis, further increasing uric acid production.

Why other options are incorrect:

- A. HGPRT: This enzyme is deficient, not increased.
- C. dATP (deoxyadenosine triphosphate): Increased dATP occurs in ADA deficiency, not Lesch-Nyhan.
- D. Branched-chain amino acids: Not related to HGPRT deficiency; elevated in maple syrup urine disease.

QUESTION

| Which of the following is true about the structural organization of proteins?

A Tertiary structure is three dimensional structure stabilized by van der waal forces

CORRECT

B Primary, secondary and tertiary structures are destroyed by denaturation

C Secondary structure is stabilized by disulphide bonds

D Secondary and Tertiary structure depends upon amino acid sequence

RIGHT ANSWER

OK **A. Tertiary structure is three dimensional structure stabilized by van der waal forces**

WHY THIS IS RIGHT

The tertiary structure of a protein is its overall 3D folding, stabilized by multiple interactions: hydrogen bonds, ionic interactions, hydrophobic interactions, and van der Waals forces. While van der Waals forces alone are weak, they contribute significantly to the compact folding of the protein.

Why other options are incorrect:

B. Primary, secondary, tertiary destroyed by denaturation: Denaturation disrupts secondary and tertiary structures, but the primary sequence remains intact.

C. Secondary structure stabilized by disulfide bonds: False; secondary structure is stabilized mainly by hydrogen bonds.

D. Secondary and tertiary structure depends upon amino acid sequence: Partially true, but this option is less precise than option A for the question's context.

QUESTION

Identify the correct statements:

1. Glucagon challenge test can be used to distinguish Von Gierke's disease and Cori's disease.
2. Choline should be avoided by patients with trimethylaminuria.
3. Oculocutaneous albinism type 1 (tyrosinase deficiency) is AR.
4. Creatine is synthesized from glycine, arginine and methionine

A 1, 2, 3, 4

CORRECT

B 1, 4

C 2, 3

D 1, 3, 4

RIGHT ANSWER

OK A. 1, 2, 3, 4

WHY THIS IS RIGHT

- Glucagon challenge test: True; it distinguishes Von Gierke's disease (G6Pase deficiency, impaired response) from Cori's disease (debranching enzyme deficiency, normal response).
- Choline in trimethylaminuria: True; dietary choline increases trimethylamine production and should be limited.
- Oculocutaneous albinism type 1: True; caused by tyrosinase deficiency and inherited in an autosomal recessive manner.
- Creatine synthesis: True; creatine is synthesized from glycine, arginine, and methionine (methionine provides methyl group via SAM).

QUESTION

Which of the following is not an amino acid-derived neurotransmitter?

A Melatonin

B Serotonin

C GABA

D **Creatine**

CORRECT

RIGHT ANSWER

OK **D. Creatine**

WHY THIS IS RIGHT

Creatine is not a neurotransmitter; it is a molecule involved in energy storage in muscles and the brain (as phosphocreatine) and does not act as a signaling molecule.

Why other options are incorrect:

A. Melatonin: Derived from tryptophan (amino acid) and acts as a neurohormone/neurotransmitter regulating circadian rhythms.

B. Serotonin: Synthesized from tryptophan and functions as a classical neurotransmitter.

C. GABA: Synthesized from glutamate (an amino acid) and is the major inhibitory neurotransmitter in the CNS.

QUESTION

A 3-month-old infant presents with liver failure, bleeding, garlic-like odor, hypophosphatemic rickets, and renal Fanconi syndrome. Which amino acid's catabolic pathway is blocked?

A Methionine

B Phenylalanine

C Tyrosine

CORRECT

D Tryptophan

RIGHT ANSWER

OK C. Tyrosine

WHY THIS IS RIGHT

- Defect: Fumarylacetoacetate hydrolase.
- This presentation is classic for Tyrosinemia type I, caused by deficiency of fumarylacetoacetate hydrolase, the final enzyme in tyrosine catabolism.
- Accumulation of toxic metabolites (fumarylacetoacetate, succinylacetone) leads to acute liver failure, bleeding diathesis, renal Fanconi syndrome, hypophosphatemic rickets, and a characteristic of garlic-like odor.
- The disease presents in infancy and is life-threatening without treatment (nitrosone, liver transplant).

Why other options are incorrect:

- A. Methionine: Causes homocystinuria, not liver failure with Fanconi syndrome.
- B. Phenylalanine: PKU causes neurodevelopmental delay, not hepatic failure.
- D. Tryptophan: Associated with Hartnup disease, mainly neurologic and skin findings.

QUESTION

Deficiency of which metal-dependent enzyme most likely explains the findings?

- A** Iron-dependent succinate dehydrogenase
- B** Copper-dependent cytochrome oxidase
- C** **Molybdenum-dependent sulfite oxidase**
- D** Manganese-dependent enolase

CORRECT

RIGHT ANSWER

- OK** **C. Molybdenum-dependent sulfite oxidase**

WHY THIS IS RIGHT

This presentation fits molybdenum cofactor deficiency, causing combined deficiency of sulfite oxidase and xanthine oxidase. Sulfite oxidase failure -> ↑ urinary sulfite with severe neonatal encephalopathy, intractable seizures, and rapid neurodegeneration. Xanthine oxidase failure -> ↑ xanthine and hypoxanthine (often with low uric acid). Thus the key impaired metal-dependent enzyme is molybdenum-dependent sulfite oxidase.

Why other options are incorrect:

- A. Iron-dependent succinate dehydrogenase: SDH defect causes mitochondrial myopathy/lactic acidosis, not sulfite/xanthine rise.
- B. Copper-dependent cytochrome oxidase: Cytochrome oxidase defects cause oxidative phosphorylation failure, not sulfite.
- D. Manganese-dependent enolase: Enolase issues affect glycolysis, not these metabolites.

QUESTION

A 15-year-old boy presents with tremors, difficulty in speech, and behavioral changes. On examination, he has hepatomegaly and a golden-brown ring at the limbus of the cornea (Kayser-Fleischer ring). Wilson's disease is suspected. Which of the following is the best investigation to support the diagnosis?

- A** Serum ceruloplasmin
- B** Liver biopsy for copper estimation
- C** Serum copper
- D** Copper excretion in urine

CORRECT

RIGHT ANSWER

- OK** **B. Liver biopsy for copper estimation**

WHY THIS IS RIGHT

Wilson disease is an autosomal recessive disorder of copper metabolism caused by defective copper transport and impaired biliary excretion. This results in excess copper accumulation in the liver, brain (basal ganglia), and cornea, producing features such as hepatomegaly, neurological symptoms, behavioral changes, and Kayser-Fleischer rings.

The most definitive investigation to support the diagnosis is liver biopsy for quantitative copper estimation, which directly measures hepatic copper concentration. Markedly increased hepatic copper confirms abnormal copper accumulation.

Why other options are incorrect:

- Serum ceruloplasmin: Often decreased in Wilson disease but can be normal in some cases, so it is not definitive.
- Serum copper: Usually low due to reduced ceruloplasmin-bound copper and is not reliable for diagnosis.
- Urinary copper excretion: Increased in Wilson disease and useful for screening/monitoring but less definitive than hepatic copper estimation.

QUESTION

A 48-year-old male who has been working in a paint manufacturing factory for over 20 years presents with chronic abdominal pain, constipation, fatigue, headache, and joint pains. He also reports irritability and sleep disturbances. On examination, pallor is noted. Laboratory investigations reveal microcytic anemia. Peripheral smear shows basophilic stippling of RBCs. Which of the following enzymes is most likely inhibited in this condition?

- A** Uroporphyrinogen III synthase
- B** Porphobilinogen synthase (ALA dehydratase)
- C** ALA synthase
- D** Ferrochelatase

CORRECT

RIGHT ANSWER

OK D. Ferrochelatase

WHY THIS IS RIGHT

Chronic exposure to lead (e.g., in paint factory workers) causes lead poisoning, which interferes with heme synthesis.

Lead inhibits two key enzymes in the heme pathway:

- Ferrochelatase -> blocks incorporation of iron into protoporphyrin IX to form heme
- ALA dehydratase (ALAD) -> inhibits conversion of ALA to porphobilinogen

Clinical features (classic):

- Abdominal pain (lead colic)
- Constipation
- Neuropathy (wrist/foot drop)
- Anemia
- Irritability, insomnia, headache

Why other options are incorrect:

- Uroporphyrinogen III synthase: Defect causes congenital erythropoietic porphyria
- Uroporphyrinogen I synthase (incorrect term): Not a standard enzyme
- ALA synthase: Rate-limiting enzyme; not inhibited by lead (actually regulated by heme)

QUESTION

A child presents with erythematous scaly patches in the perioral region, mucosal ulcers, and impaired epithelial wound healing. What is the likely mineral deficiency associated with this condition?

A Iron deficiency

B Zinc deficiency

CORRECT

C Calcium deficiency

D Copper deficiency

RIGHT ANSWER

B. Zinc deficiency

WHY THIS IS RIGHT

Zinc deficiency leads to characteristic periorificial dermatitis (around mouth, eyes, anus), mucosal ulcers, and impaired wound healing, as seen in this child. Zinc is a cofactor for numerous enzymes involved in:

- DNA & RNA synthesis
- Cell division and tissue repair
- Protein synthesis and epithelial integrity

Deficiency results in: Acrodermatitis enteropathica (genetic zinc malabsorption)

- Impaired epithelial regeneration -> erythematous scaly lesions
- Poor wound healing
- Mucosal breakdown -> ulcers

Other features of zinc deficiency:

- Alopecia
- Diarrhea
- Growth retardation
- Loss of taste (hypogeusia)

Why other options are incorrect:

- Iron deficiency: Causes anemia, not characteristic dermatitis
- Calcium deficiency: Affects bones (rickets/osteomalacia)
- Copper deficiency: Causes anemia and neurologic issues, not perioral dermatitis

QUESTION

A 2-year-old child is brought with progressive pallor, recurrent infections, and failure to thrive. On examination, the child has hypopigmented skin and sparse, light-colored hair. Laboratory investigations reveal anemia and neutropenia. Which of the following micronutrient deficiencies best explains this clinical presentation?

A Zinc

B Selenium

C Iron

D Copper

CORRECT

RIGHT ANSWER

OK

D. Copper

WHY THIS IS RIGHT

The combination of anemia + neutropenia + hypopigmentation of skin and hair is characteristic of copper deficiency.

Copper is a cofactor for several important enzymes:

- Ceruloplasmin -> involved in iron metabolism ($\text{Fe}^{2+} \rightarrow \text{Fe}^{3+}$)
- Cytochrome c oxidase -> energy production
- Tyrosinase -> melanin synthesis

Pathophysiology:

- ↓ Ceruloplasmin -> impaired iron mobilization -> anemia
- Impaired immune function -> neutropenia
- ↓ Tyrosinase activity -> ↓ melanin -> hypopigmentation

Clinical associations:

- Malnutrition
- Excess zinc intake (interferes with copper absorption)
- Menkes disease (genetic copper deficiency)

Why other options are incorrect:

- Zinc: Causes dermatitis, delayed wound healing
- Selenium: Cardiomyopathy (Keshan disease)
- Iron: Causes anemia only (no neutropenia or hypopigmentation)

QUESTION

A patient's relatives sent a message on social media to the consulting doctor, unaware about the other investigations. But he mentioned that urinary coproporphyrin is positive. What is the probable cause?

A Lead poisoning

CORRECT

B Asbestosis

C Silicosis

D Mercury poisoning

RIGHT ANSWER

OK **A. Lead poisoning**

WHY THIS IS RIGHT

Lead poisoning interferes with heme synthesis, leading to accumulation of porphyrin precursors such as coproporphyrin in urine. Lead inhibits key enzymes in the heme pathway:

- ALA dehydratase (ALAD)
- Ferrochelatase

Consequences:

- ↑ ALA and coproporphyrin in urine
- ↓ heme synthesis → anemia

Clinical features:

- Abdominal pain (lead colic)
- Neurological symptoms (irritability, neuropathy)
- Anemia with basophilic stippling
- Wrist/foot drop

Why other options are incorrect:

- Asbestosis: Lung fibrosis (no effect on heme synthesis)
- Silicosis: Occupational lung disease
- Mercury poisoning: Neurological and renal toxicity, not porphyrin metabolism

QUESTION

Heavy metal chelators are all EXCEPT:

A Penicillamine

B Oxime

CORRECT

C BAL

D Trientine

RIGHT ANSWER

OK B. Oxime

WHY THIS IS RIGHT

Heavy metal chelators are agents that bind toxic metals and form stable complexes that can be excreted from the body, thereby reducing metal toxicity.

Examples include:

- Penicillamine - used in copper poisoning and Wilson disease.
- BAL (Dimercaprol) - used in arsenic, mercury, and gold poisoning.
- Trientine - another copper chelator used in Wilson disease.

Oximes (e.g., pralidoxime) are not heavy-metal chelators. They are used in organophosphate poisoning to reactivate acetylcholinesterase by removing the phosphate group from the inhibited enzyme.

QUESTION

Which of the following connective tissue degradation disorder is associated with aortic aneurysm?

A Ehlers-Danlos Syndrome

B Marfan's Syndrome

CORRECT

C Atherosclerosis

D Scurvy

RIGHT ANSWER

B. Marfan's Syndrome

WHY THIS IS RIGHT

Marfan syndrome is a connective tissue disorder caused by mutation in the fibrillin-1 (FBN1) gene, leading to defective elastic fiber formation in connective tissues. Weakness of the aortic media predisposes to aortic root dilatation, aneurysm, and dissection, which is a major cause of mortality in these patients.

It is also characterized by tall stature, long limbs (arachnodactyly), lens dislocation, and skeletal abnormalities.

Why not other options:

- Ehlers-Danlos syndrome: Defect in collagen synthesis causing hyperextensible skin and joint hypermobility; vascular complications may occur in rare types but aortic aneurysm classically associated with Marfan syndrome.
- Atherosclerosis: Causes arterial narrowing and plaque formation, not primarily a connective-tissue degradation disorder.
- Scurvy: Vitamin C deficiency causing defective collagen synthesis leading to bleeding gums and poor wound healing, not aortic aneurysm.

QUESTION

Which of the following deficiencies is associated with sexual maturation defect and hypogonadism?

A Selenium

B Zn

CORRECT

C Cu

D Iron

RIGHT ANSWER

OK B. Zn

WHY THIS IS RIGHT

Zinc deficiency is associated with delayed sexual maturation, hypogonadism, and impaired reproductive function. Zinc plays an important role in DNA synthesis, cell division, and hormone metabolism, including the normal function of the hypothalamic-pituitary-gonadal axis.

Deficiency of zinc can lead to delayed puberty, impaired spermatogenesis, decreased testosterone levels, growth retardation, poor wound healing, and decreased immunity.

Therefore, zinc is essential for normal sexual development and reproductive health, and its deficiency can result in hypogonadism and delayed sexual maturation.

QUESTION

A Patient had dietary Cu deficiency, which is required by lysyl oxidase to cross link the collagen, what is the diagnosis of the patient?

A Menkes Disease

CORRECT

B Scoliosis

C Ehlers Danlos syndrome

D Scurvy

RIGHT ANSWER

A. Menkes Disease

WHY THIS IS RIGHT

Copper is an essential cofactor for the enzyme lysyl oxidase, which catalyzes the oxidative deamination of lysine and hydroxylysine residues in collagen and elastin, allowing cross-linking of connective tissue fibers. Role of lysyl oxidase:

- Requires Cu^{2+} (copper) as a cofactor
- Forms cross-links between collagen and elastin fibers
- Provides tensile strength and structural stability to connective tissue

Menkes disease:

- X-linked recessive disorder
- Caused by mutation in ATP7A gene -> defective copper transport
- Leads to systemic copper deficiency and reduced lysyl oxidase activity
- Weak connective tissue
- Kinky (steely) hair
- Neurodegeneration
- Failure to thrive
- Hypopigmentation

Differentiation from other options:

- Scurvy -> Vitamin C deficiency -> impaired hydroxylation of collagen
- Ehlers-Danlos syndrome -> collagen structure/synthesis defects
- Scoliosis -> spinal curvature disorder, not enzyme-related

QUESTION

Which of the following is false regarding jaundice?

- A** Urobilinogen is usually increased in hemolytic jaundice
- B** Urine bilirubin is absent in hemolytic jaundice.
- C** Crigler-Najjar syndrome presents with unconjugated hyperbilirubinemia
- D** Obstructive jaundice shows a negative Van der Bergh reaction

CORRECT

RIGHT ANSWER

- OK** **D. Obstructive jaundice shows a negative Van der Bergh reaction**

WHY THIS IS RIGHT

The Van der Bergh reaction detects conjugated bilirubin. In obstructive (post-hepatic) jaundice, conjugated bilirubin accumulates in blood and urine, giving a positive Van der Bergh reaction. A negative reaction would be incorrect.

Why the others are correct:

- A. Urobilinogen increased in hemolytic jaundice: Hemolysis increases unconjugated bilirubin, which is partially converted to urobilinogen in the intestine.
- B. Urine bilirubin absent in hemolytic jaundice: Unconjugated bilirubin is not water-soluble, so it is not excreted in urine.
- C. Crigler-Najjar syndrome: Causes unconjugated hyperbilirubinemia due to UDP-glucuronyl transferase deficiency.

QUESTION

Cells that are exposed to longer ischemic periods are found to have reduced numbers of ribosomes attached to the endoplasmic reticulum. This structural change is most likely to impair which of the following cellular functions?

A Drug detoxification

B Synthesis of cell membrane proteins

CORRECT

C Synthesis of cytosolic proteins

D Synthesis of steroid hormones

RIGHT ANSWER

OK

B. Synthesis of cell membrane proteins

WHY THIS IS RIGHT

Ribosomes attached to the rough endoplasmic reticulum (RER) synthesize secretory and membrane-bound proteins. Reduced ribosome attachment during ischemia impairs the production of integral membrane proteins and secreted proteins, disrupting normal cellular function and signaling.

Why the others are incorrect:

A. Drug detoxification: Mainly occurs via smooth ER enzymes (e.g., cytochrome P450), independent of ribosome attachment.

C. Synthesis of cytosolic proteins: Carried out by free ribosomes in the cytosol, not affected by RER ribosome reduction.

D. Synthesis of steroid hormones: Occurs in the smooth ER, which does not require ribosome attachment.

QUESTION

A 62-year-old man comes to the OPD due to an enlarging mole on his right forearm. Excisional biopsy is performed and histopathology reveals malignant melanoma. Immunohistochemical analysis indicates that the malignant cells have decreased integrin expression. These cells are most likely to exhibit poor adhesion to which of the following components of the extracellular matrix?

- A** Actin
- B** Fibronectin
- C** Hyaluronic acid
- D** Keratan sulfate

CORRECT

RIGHT ANSWER

B. Fibronectin

WHY THIS IS RIGHT

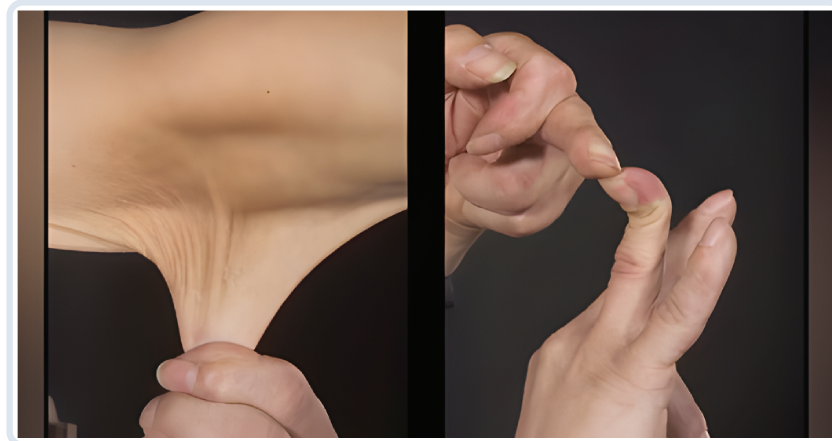
Integrins are transmembrane receptors that mediate cell adhesion to extracellular matrix (ECM) proteins such as fibronectin, laminin, and collagen. Decreased integrin expression in melanoma cells reduces adhesion to fibronectin, facilitating detachment, invasion, and metastasis.

Why the others are incorrect:

- A. Actin: Cytoskeletal protein inside the cell; integrins connect to actin, but actin itself is not an ECM component.
- C. Hyaluronic acid: A glycosaminoglycan in ECM; adhesion involves CD44, not integrins.
- D. Keratan sulfate: ECM proteoglycan; not a primary ligand for integrins in melanoma cell adhesion.

QUESTION

24-year-old woman comes to the OPD for a preemployment medical evaluation. The patient has no known medical problems but reports that her skin bruises and scars easily. She says that most of her family members have a very "flexible" body, and her brother works in a circus as a contortionist. Physical examination findings are shown. This patient most likely has an inherited defect in which of the following proteins?



A Collagen

CORRECT

B Elastin

C Fibrillin-1

D Hyaluronic acid

RIGHT ANSWER

OK A. Collagen

WHY THIS IS RIGHT

This patient likely has Ehlers-Danlos syndrome (EDS), suggested by hyperextensible skin, easy bruising, and family history of joint flexibility. EDS is caused by defects in collagen synthesis or structure, leading to fragile connective tissue, skin hyperelasticity, and joint hypermobility.

Why the others are incorrect:

B. Elastin: Defects cause vascular abnormalities and localized skin laxity, not systemic hypermobility with easy bruising.

C. Fibrillin-1: Mutations cause Marfan syndrome, which features tall stature, aortic aneurysms, and lens dislocation rather than pronounced skin fragility.

D. Hyaluronic acid: Structural ECM component; deficiencies do not produce EDS-like manifestations.

QUESTION

Selenocysteine is present in which of the following enzymes?

1. Glutathione reductase
2. Glutathione peroxidase
3. Thioredoxine peroxidase
4. Thioredoxine reductase
5. Iodothyronine deiodinase

A 1, 2, 4

B 2, 4, 5

CORRECT

C 1, 3, 5

D 1, 2, 3, 4, 5

RIGHT ANSWER

OK B. 2, 4, 5

WHY THIS IS RIGHT

Selenocysteine is the 21st amino acid, incorporated into enzymes that require selenium for redox activity. Among the listed enzymes:

- Glutathione peroxidase (2): Contains selenocysteine at the active site, catalyzing reduction of peroxides.
- Thioredoxin reductase (4): Uses selenocysteine to maintain redox homeostasis.
- Iodothyronine deiodinase (5): Contains selenocysteine for thyroid hormone activation/inactivation.

Why other options are incorrect:

- Glutathione reductase (1): Uses FAD, does not contain selenocysteine.
- Thioredoxin peroxidase (3): Typically uses cysteine, not selenocysteine.

QUESTION

| Which of the following is true about Hemochromatosis?

- A** The mRNA for Transferrin receptor is stabilised by binding of IRP to 3' stem-loop structures known as IRE.
- B** The mRNA for the Transferrin receptor is not bound by IRP and is rapidly degraded. CORRECT
- C** The mRNA for apoferritin is not bound by IRP at 5' stem loop IRE and is rapidly degraded.
- D** The mRNA for apoferritin is bound by IRP and is not translated.

RIGHT ANSWER

- OK** **B. The mRNA for the Transferrin receptor is not bound by IRP and is rapidly degraded.**

WHY THIS IS RIGHT

In hemochromatosis, there is iron overload, so cellular iron levels are high. Under these conditions, iron regulatory proteins (IRPs) cannot bind to the iron-responsive elements (IREs) on the 3' UTR of transferrin receptor mRNA, leading to rapid degradation. This decreases transferrin receptor synthesis, reducing iron uptake—a protective response to high intracellular iron.

Why other options are incorrect:

A. The mRNA for Transferrin receptor is stabilised by binding of IRP to 3' stem-loop structures known as IRE: Occurs only during iron deficiency, not overload.

C. The mRNA for apoferritin is not bound by IRP at 5' stem loop IRE and is rapidly degraded and D. The mRNA for apoferritin is bound by IRP and is not translated: IRP binds the 5' IRE of ferritin mRNA when iron is low to block translation; during iron overload, ferritin is translated, so these statements are false.

QUESTION

Which of the following enzymes is produced in increased amounts than normal in the liver of a patient with lead toxicity?

A ALA synthase

CORRECT

B Heme oxygenase

C Ferrochelatase

D Porphobilinogen deaminase

RIGHT ANSWER

OK A. ALA synthase

WHY THIS IS RIGHT

In lead poisoning, lead inhibits ferrochelatase and ALA dehydratase, leading to a decreased production of heme. As a compensatory response, the liver upregulates ALA synthase, the rate-limiting enzyme of heme synthesis, to try to restore heme levels. This results in accumulation of ALA and protoporphyrin, causing the biochemical and clinical manifestations of lead toxicity.

Why other options are incorrect:

B. Heme oxygenase: Breaks down heme; not induced by lead toxicity.

C. Ferrochelatase: Directly inhibited by lead, so activity decreases.

D. Porphobilinogen deaminase: Activity is not increased in lead poisoning; defects in this enzyme cause acute intermittent porphyria, not lead toxicity.

QUESTION

Which component of fiber delays gastric emptying and reduces cholesterol absorption?

A Pectin

CORRECT

B Cellulose

C Hemicellulose

D Lignin

RIGHT ANSWER

OK **A. Pectin**

WHY THIS IS RIGHT

Pectin is a soluble dietary fiber that forms a viscous gel in the gastrointestinal tract. This gel delays gastric emptying, slows intestinal transit, and binds bile acids, leading to increased bile excretion and reduced cholesterol absorption. These properties help lower LDL cholesterol and improve postprandial glycemic control.

Why other options are incorrect:

- B. Cellulose: An insoluble fiber that mainly increases stool bulk without affecting cholesterol absorption.
- C. Hemicellulose: Primarily insoluble with limited effect on gastric emptying or bile binding.
- D. Lignin: Insoluble and non-fermentable; does not significantly reduce cholesterol absorption.

QUESTION

A patient with severe malnutrition presents with muscle pain, proximal muscle weakness, and cardiomyopathy. Labs show markedly decreased activity of an enzyme that converts $\text{H}_2\text{O}_2 \rightarrow \text{H}_2\text{O}$ using glutathione as a cofactor. Which trace element deficiency is most likely responsible?

A Zinc

B Copper

C Selenium

CORRECT

D Manganese

RIGHT ANSWER

OK

C. Selenium

WHY THIS IS RIGHT

The enzyme converting $\text{H}_2\text{O}_2 \rightarrow \text{H}_2\text{O}$ using glutathione is glutathione peroxidase, a selenium-dependent enzyme. Selenium deficiency leads to impaired detoxification of reactive oxygen species, causing oxidative muscle damage, proximal myopathy, and dilated cardiomyopathy (classically seen in severe malnutrition, e.g., Keshan disease). Reduced glutathione peroxidase activity explains the accumulation of hydrogen peroxide and resultant tissue injury.

Why other options are incorrect:

- A. Zinc: Required for many enzymes and transcription factors, not glutathione peroxidase.
- B. Copper: Cofactor for cytochrome c oxidase and SOD, not H_2O_2 detoxification via glutathione.
- D. Manganese: Cofactor for mitochondrial superoxide dismutase, not glutathione peroxidase.

QUESTION

| All are tests for bile pigment except:

A Fouchet's test

B Gmelin's test

C Harrison's test

D Hay's sulfur test

CORRECT

RIGHT ANSWER

OK **D. Hay's sulfur test**

WHY THIS IS RIGHT

Tests for bile pigments (bilirubin):

- Fouchet's test -> Bilirubin -> green color
- Gmelin's test -> Rainbow colors due to oxidation of bile pigments
- Harrison's test -> Test for bile pigments in urine
- Hammarsten test (extra) -> Also for bile pigments

QUESTION

A newborn presents with intractable seizures, severe neurological deterioration, and elevated sulfite levels in urine. Xanthine and hypoxanthine are also markedly increased.

Deficiency of which metal-dependent enzyme MOST likely explains the findings?

A Iron-dependent succinate dehydrogenase

B Copper-dependent cytochrome oxidase

C Molybdenum-dependent sulfite oxidase

CORRECT

D Manganese-dependent enolase

RIGHT ANSWER

OK C. Molybdenum-dependent sulfite oxidase

WHY THIS IS RIGHT

This presentation fits molybdenum cofactor deficiency, causing combined deficiency of sulfite oxidase and xanthine oxidase. Sulfite oxidase failure -> ↑ urinary sulfite with severe neonatal encephalopathy, intractable seizures, and rapid neurodegeneration. Xanthine oxidase failure -> ↑ xanthine and hypoxanthine (often with low uric acid). Thus the key impaired metal-dependent enzyme is molybdenum-dependent sulfite oxidase.

Why other options are incorrect:

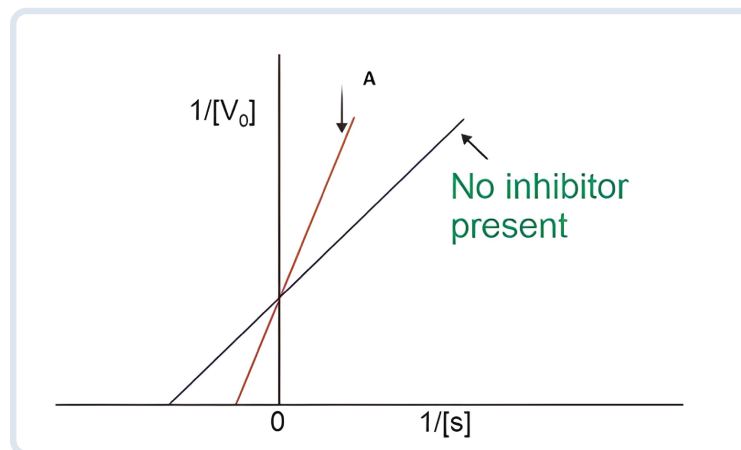
A: SDH defect causes mitochondrial myopathy/lactic acidosis, not sulfite/xanthine rise.

B: Cytochrome oxidase defects cause oxidative phosphorylation failure, not sulfite.

D: Enolase issues affect glycolysis, not these metabolites.

QUESTION

Identify the type of inhibition exhibited by A.



CORRECT

A Competitive inhibition

B Uncompetitive inhibition

C Mixed inhibition

D Non-competitive inhibition

RIGHT ANSWER

OK A. Competitive inhibition

WHY THIS IS RIGHT

The graph represents competitive inhibition, where the inhibitor competes with the substrate for the active site of the enzyme.

Key features in Lineweaver-Burk plot:

- Same Y-intercept ($1/V_{max}$ unchanged)
- Slope increases
- X-intercept shifts toward zero ($\uparrow K_m$)

Interpretation:

- V_{max} remains unchanged $\rightarrow$ inhibition can be overcome by increasing substrate concentration
- K_m increases $\rightarrow$ decreased affinity of enzyme for substrate (more substrate needed)

Why other options are incorrect:

- Uncompetitive inhibition: Parallel lines ($\downarrow K_m$ and $\downarrow V_{max}$)
- Non-competitive inhibition: $\downarrow V_{max}$, K_m unchanged (same X-intercept)
- Mixed inhibition: Both K_m and V_{max} change (lines intersect off axes)

QUESTION

Detoxification of Xenobiotics for phase 1 reaction (Hydroxylation) require monooxygenase/cytochrome P-450 along with:

A NADPH

CORRECT

B NADH

C FAD

D FADH₂

RIGHT ANSWER

OK

A. NADPH

WHY THIS IS RIGHT

Phase I detoxification (functionalization reactions) of xenobiotics mainly involves oxidation reactions such as hydroxylation, catalyzed by the cytochrome P450 monooxygenase system present in the smooth endoplasmic reticulum of liver cells.

In this system, NADPH acts as the reducing agent and provides electrons through NADPH-cytochrome P450 reductase, which allows molecular oxygen to be split, with one oxygen atom incorporated into the substrate (hydroxylation) and the other reduced to water.

Why other options are incorrect:

- NADH: Mainly involved in mitochondrial electron transport and not the primary reducing agent for cytochrome P450 reactions.
- FAD / FADH₂: These act as prosthetic groups in some enzymes but are not the direct electron donors for the cytochrome P450 monooxygenase system.

QUESTION

The active site of enzyme is complimentary to:

A Transition state

B Allosteric activator

C **Substrate**

CORRECT

D Product

RIGHT ANSWER

OK **C. Substrate**

WHY THIS IS RIGHT

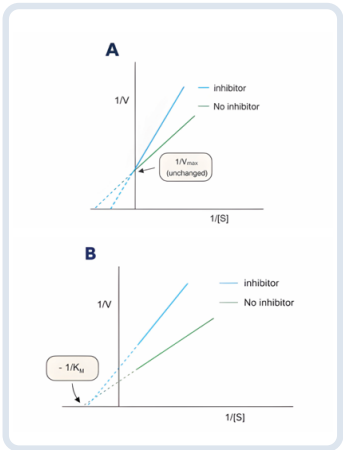
The active site of an enzyme is a specific region where the substrate binds to form the enzyme-substrate complex. The shape and chemical properties of the active site are complementary to the substrate, allowing specific recognition and binding, which enables the enzyme to catalyze the reaction efficiently.

Why other options are incorrect:

- Transition state: The enzyme stabilizes the transition state during catalysis, but the active site is primarily designed to bind the substrate initially.
- Allosteric activator: These molecules bind to allosteric sites, not the active site.
- Product: Products are formed after the reaction and typically have lower affinity for the active site, allowing their release.

QUESTION

Inhibitor A and B are



- A

Competitive and non-competitive

CORRECT
- B

Uncompetitive and non-competitive
- C

Non-competitive and uncompetitive
- D

Non-competitive and competitive
- OK

RIGHT ANSWER

A. Competitive and non-competitive

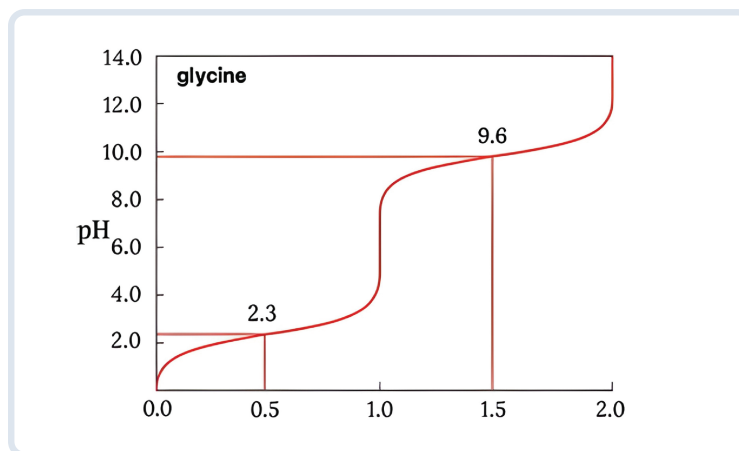
WHY THIS IS RIGHT

Inhibitor A is a competitive inhibitor, and Inhibitor B is a non-competitive inhibitor.

Type of inhibition	Constant Parameter	Variable Parameter	Lineweaver-Burk Plot Intersection
Competitive Inhibition	V_{max} (unchanged)	K_m (increased) V_{max}	On Y-axis (same V_{max})
Non-Competitive Inhibition	K_m (unchanged)	(decreased)	On X-axis (same K_m)

QUESTION

In the given glycine titration curve, calculate or identify the pH at which glycine predominantly exists in its zwitterionic form?



A 4.4

B 6

CORRECT

C 9

D 2.2

RIGHT ANSWER

OK

B. 6

WHY THIS IS RIGHT

Glycine's $pI = (pK_a 2.3 + pK_a 9.6) \div 2 \approx 6$, where it exists mainly as a zwitterion with zero net charge.

At isoelectric pH:

- Proteins have no electrophoretic mobility.
- Minimum solubility -> maximal precipitation.
- Least stability, prone to aggregation.
- Do not bind to ion-exchange columns.

QUESTION

| When $pK_a = pH$

CORRECT

- A** Conc. Of drug is 50% ionic and 50% non ionic
- B** Absorption of drug is 50% in ionic form and 50% non ionic
- C** Conc. of drug is 75% ionic and 25% non ionic
- D** Conc. of drug is 25% ionic and 75% non ionic

RIGHT ANSWER

- OK** **A. Conc. Of drug is 50% ionic and 50% non ionic**

WHY THIS IS RIGHT

According to the Henderson-Hasselbalch equation, when $pH = pK_a$, the ratio of ionized to unionized drug is 1:1. This means 50% of the drug exists in its ionic form and 50% in its non-ionic (unionized) form, which is critical for drug absorption since unionized drugs cross membranes more easily.

Why the others are incorrect:

B. 50% absorption in ionic and non-ionic form: Absorption depends on unionized fraction, not directly 50/50.

C. 75% ionic / 25% non-ionic or D. 25% ionic / 75% non-ionic: These occur only when pH differs from pK_a by ± 0.5 -1 units, not when $pH = pK_a$.

QUESTION

A 42-year-old man is brought to the emergency department due to nausea, vomiting, and blurred vision. He was making his own whiskey with a home moonshine still and started feeling sick after sampling the first several ounces of distillate. Funduscopic evaluation reveals optic disc hyperemia. Laboratory results show a high anion gap metabolic acidosis. He is started on fomepizole, a medication that transiently binds to alcohol dehydrogenase. Which of the following graphs most accurately portrays the change in enzyme kinetics after administration of the antidote?

CORRECT

A

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B

https://corebtr-assets-production.s3.ap-south-1.amazonaws.com/questions_image/option/01KN3NNW63GWX9RPRFYECXHR74.jpg

C

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D

https://corebtr-assets-production.s3.ap-south-1.amazonaws.com/questions_image/option/01KN3NNWDE9VCSW72CBWT4QNW.jpg

RIGHT ANSWER

OK

A. https://corebtr-assets-production.s3.ap-south-1.amazonaws.com/questions_image/option/01KN3NNW156RCG1XTPEP0RPZFM.jpg

WHY THIS IS RIGHT

This patient has methanol poisoning, suggested by homemade alcohol ingestion, nausea, vomiting, blurred vision, optic disc hyperemia, and high anion gap metabolic acidosis. Fomepizole acts as a competitive inhibitor of alcohol dehydrogenase, the enzyme that converts methanol to toxic formaldehyde. In competitive inhibition, the apparent K_m increases (substrate affinity decreases), but V_{max} remains unchanged. Therefore, the correct graph shows a rightward shift of the Michaelis-Menten curve without change in maximum velocity.

Why the others are incorrect:

- Graphs showing decreased V_{max} represent noncompetitive inhibition, not applicable here.
- Graphs showing unchanged K_m but decreased V_{max} do not reflect competitive binding of fomepizole.

QUESTION

A 22-year-old man comes to the office due to recurrent blistering on the back of his hands and forearms for the past several years as shown. The patient usually develops small itchy spots but lately has had large blisters that heal with hyperpigmentation after rupturing. The patient works as a night security guard and has had no exposure to chemicals or animals. He drinks 2-3 cans of beer daily. Which of the following enzymes is most likely deficient in this patient?



A Aminolevulinate dehydratase

B Aminolevulinate synthase

C Porphobilinogen deaminase

D Uroporphyrinogen decarboxylase

CORRECT

RIGHT ANSWER

OK D. Uroporphyrinogen decarboxylase

WHY THIS IS RIGHT

This patient has porphyria cutanea tarda (PCT), indicated by chronic blistering on sun-exposed areas, hyperpigmentation, and alcohol use as a trigger. PCT results from deficiency of uroporphyrinogen decarboxylase, leading to accumulation of uroporphyrins in the skin, causing photosensitivity and blistering.

Why the others are incorrect:

A. Aminolevulinate dehydratase: Deficiency causes ALA dehydratase porphyria, a rare acute porphyria without skin involvement.

B. Aminolevulinate synthase: Rate-limiting enzyme; defects typically cause X-linked sideroblastic anemia, not PCT.

C. Porphobilinogen deaminase: Deficiency causes acute intermittent porphyria, which presents with neurovisceral symptoms, not chronic skin blistering.

QUESTION

28-year-old woman comes to the emergency department with acute-onset abdominal pain, nausea, and confusion. She has no significant past medical history and does not use tobacco or alcohol as they have made her feel sick in the past. A sample of her urine is reddish in color and darkens on standing for 24 hours. Intravenous dextrose is administered, and her symptoms improve significantly. Dextrose infusion most likely improved this patient's condition by affecting which of the following pathways?

- A** Gluconeogenesis
- B** Fatty-acid-synthesis
- C** Ketone-body formation
- D** Porphyrin synthesis

CORRECT

RIGHT ANSWER

- OK** **D. Porphyrin synthesis**

WHY THIS IS RIGHT

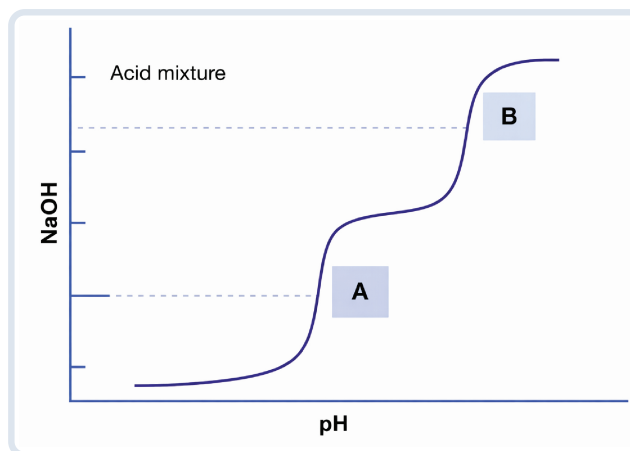
This patient presents with acute intermittent porphyria (AIP), suggested by acute abdominal pain, neuropsychiatric symptoms, reddish urine that darkens on standing, and symptom improvement with intravenous dextrose. Glucose administration suppresses ALA synthase, the rate-limiting enzyme in hepatic porphyrin synthesis, reducing accumulation of neurotoxic porphyrin precursors and alleviating symptoms.

Why the others are incorrect:

- A. Gluconeogenesis: Dextrose provides glucose but does not directly target gluconeogenesis in this acute setting.
- B. Fatty-acid synthesis: Not involved in AIP pathophysiology.
- C. Ketone-body formation: Relevant in fasting/starvation, not porphyria management.

QUESTION

The graph shown below is the titration curve of a biochemical compound. Which of the following statement is true?



A The maximum buffering capacity of the compound is represented by points A and B

CORRECT

B The points A and B represent the range of maximum ionization of the amine and carboxyl group

C The compound has three ionisable side chains

D The compound has one ionisable group

RIGHT ANSWER

OK A. The maximum buffering capacity of the compound is represented by points A and B

WHY THIS IS RIGHT

In a titration curve of an amino acid or similar biochemical compound:

- Points A and B correspond to the pKa values of ionizable groups (e.g., carboxyl and amino groups).
- The flattest regions of the curve indicate maximum buffering capacity, where the compound can resist changes in pH effectively.
- For a simple amino acid with two ionizable groups, these two regions are typically observed around the pKa of the carboxyl group (~ 2) and the pKa of the amino group ($\sim 9-10$).

Why other options are incorrect:

- B. A and B represent the range of maximum ionization: Incorrect; they indicate buffering regions, not the full ionization range.
- C. Three ionizable side chains: Would show three flat buffering regions, not just A and B.
- D. One ionizable group: Would only have one buffering region, not two.

QUESTION

| Which is correct about isoenzymes?

- A** Same action with similar kinetics
- B** Same quaternary structure
- C** Catalyse the same reaction. CORRECT
- D** Cannot be separated from each other on Electrophoresis.

RIGHT ANSWER

- OK** **C. Catalyse the same reaction.**

WHY THIS IS RIGHT

Isoenzymes (isozymes) are different molecular forms of an enzyme that catalyze the same chemical reaction but may differ in amino acid sequence, kinetic properties, regulatory features, or tissue distribution. This allows fine-tuning of metabolism in different tissues or under different physiological conditions.

Why other options are incorrect:

- A. Same action with similar kinetics: False; isoenzymes often have different K_m , V_{max} , or regulatory properties.
- B. Same quaternary structure: False; isoenzymes may have different subunit composition.
- D. Cannot be separated on electrophoresis: False; isoenzymes can often be separated based on charge or size using electrophoresis.

QUESTION

Succinate dehydrogenase (SDH) is an enzyme complex located within the inner mitochondrial membrane that catalyses the oxidation of succinate to fumarate. An experiment is conducted to determine if malate alters the rate of SDH activity. Reaction velocity is measured with and without a fixed quantity of malate as succinate concentration is gradually increased. Which of the following is the most accurate statement about malate in this experiment?

Succinate concentration (mM)	Rate of reaction without malate ($\mu\text{mol} / \text{L} / \text{sec}$)	Rate of reaction with malate ($\mu\text{mol} / \text{L} / \text{sec}$)
2	80	40
8	200	120
16	280	200
64	400	400
128	400	400

- A** It alters the maximal velocity of the reaction
- B** It binds the enzyme at a different site than succinate
- C** It decreases affinity of the enzyme for succinate

D It is a competitive inhibitor of the enzyme

CORRECT

RIGHT ANSWER

OK D. It is a competitive inhibitor of the enzyme

WHY THIS IS RIGHT

In this experiment, malate competes with succinate for binding to succinate dehydrogenase (SDH). As a structural analog of succinate, malate binds the active site, preventing succinate from binding, which is the hallmark of competitive inhibition. This is why, as succinate concentration increases, the inhibition can be overcome, and the reaction velocity eventually reaches the normal V_{max} .

Why other options are incorrect:

- A. It alters the maximal velocity of reaction: V_{max} is unchanged in competitive inhibition.
- B. It binds the enzyme at a different site than succinate: False; it binds the same active site.
- C. It decreases affinity of the enzyme for succinate: While apparent K_m increases, the key mechanism is competition, not allosteric change.

QUESTION

Which of the following is not used for protein precipitation?

- A** Heavy metals
- B** Organic solvents
- C** Change in pH other than isoelectric pH
- D** Trichloroacetic acid

CORRECT

RIGHT ANSWER

- OK** **C. Change in pH other than isoelectric pH**

WHY THIS IS RIGHT

Protein precipitation occurs most efficiently at the isoelectric pH (pI), where the net charge on the protein is zero, minimizing electrostatic repulsion and promoting aggregation. Changing the pH away from the isoelectric point increases the net charge on proteins, enhancing solubility rather than causing precipitation. Hence, pH changes other than pI are not useful for protein precipitation.

Why other options are incorrect:

- A. Heavy metals: Form insoluble complexes with proteins, causing precipitation.
- B. Organic solvents: Reduce hydration shell and dielectric constant, leading to precipitation.
- D. Trichloroacetic acid: A strong acid commonly used to precipitate proteins by denaturation.

QUESTION

| Gel filtration chromatography separates molecules mainly based on:

A Polarity

B Charge

C Size

CORRECT

D pKa

RIGHT ANSWER

OK C. Size

WHY THIS IS RIGHT

- Large molecules elute first.
- Gel filtration chromatography (also called Size Exclusion Chromatography) separates molecules purely based on molecular size, not charge or polarity.
- The column contains porous beads:

1. Large molecules cannot enter the pores -> they take a shorter path -> elute first.

2. Small molecules enter deep into the pores -> get delayed -> elute last.

-> Separation = inverse of size

-> Big = fast

-> Small = slow

Why other options are incorrect:

A. Polarity: Separation by polarity is characteristic of normal-phase chromatography.

B. Charge: Ion-exchange chromatography separates molecules based on charge.

D. pKa: pKa influences ionization, not size-exclusion separation.

QUESTION

A patient overdoses on a heavy metal. The toxin binds to an enzyme at a site distinct from the active site, producing a noncompetitive inhibition pattern. Which treatment is most appropriate?

A Increase substrate concentration

B Add a chelating agent

CORRECT

C Administer a substrate analog

D Give excess cofactor

RIGHT ANSWER

OK **B. Add a chelating agent**

WHY THIS IS RIGHT

Heavy metals (e.g., lead, mercury) often cause noncompetitive inhibition by binding to an allosteric site or sulfhydryl groups on enzymes, reducing V_{max} without changing K_m . Because the inhibitor does not compete with the substrate at the active site, increasing substrate concentration does not overcome inhibition. The most effective treatment is a chelating agent (e.g., EDTA, dimercaprol), which binds the heavy metal and facilitates its removal, restoring enzyme activity.

Why other options are incorrect:

A. Increase substrate concentration: Increasing substrate cannot overcome noncompetitive inhibition.

C. Administer substrate analog: Substrate analogs act as competitive inhibitors, worsening inhibition.

D. Give excess cofactor: Excess cofactor does not displace heavy metals from allosteric binding sites.

QUESTION

A family that consumes a diet predominantly consisting of polished rice develops symptoms of muscle weakness and difficulty in movement. Which of the following nutrients should be administered as prophylaxis?

A Calcium

B Thiamine

CORRECT

C Vitamin D

D Iron

RIGHT ANSWER

OK B. Thiamine

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$19” here and does not include a written explanation for this item.

QUESTION

A 45-year-old woman presents with fatigue, pallor, and glossitis. Laboratory evaluation reveals macrocytic anemia. Serum studies show elevated homocysteine levels, while methylmalonic acid (MMA) levels are normal. Which of the following is the most likely underlying cause?

A Vitamin B12 deficiency

B Folate deficiency

CORRECT

C Iron deficiency anemia

D Thalassemia minor

RIGHT ANSWER

OK B. Folate deficiency

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$1a” here and does not include a written explanation for this item.

QUESTION

Which of the following vitamin deficiencies is most likely to cause lactic acidosis?

A Niacin

B Vitamin C

C Thiamine

CORRECT

D Riboflavin

RIGHT ANSWER

OK C. Thiamine

WHY THIS IS RIGHT

Thiamine (Vitamin B1) is an essential cofactor for several enzymes involved in aerobic carbohydrate metabolism, particularly:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase

When thiamine is deficient, the pyruvate dehydrogenase reaction is impaired, preventing the conversion of pyruvate to acetyl-CoA. As a result, pyruvate is instead converted to lactate, leading to accumulation of lactic acid and lactic acidosis.

Why other options are incorrect:

- Niacin: Deficiency causes pellagra (dermatitis, diarrhea, dementia).
- Vitamin C: Deficiency leads to scurvy due to impaired collagen synthesis.
- Riboflavin: Deficiency causes cheilosis, glossitis, and dermatitis, not lactic acidosis.

QUESTION

A woman on anti-depressants presents with bleeding. She gives a history of bulky stools which sticks to the pan. Which of the following vitamin deficiency can cause bleeding in this condition?

A Vitamin A

B Vitamin D

C Vitamin E

D Vitamin K

CORRECT

RIGHT ANSWER

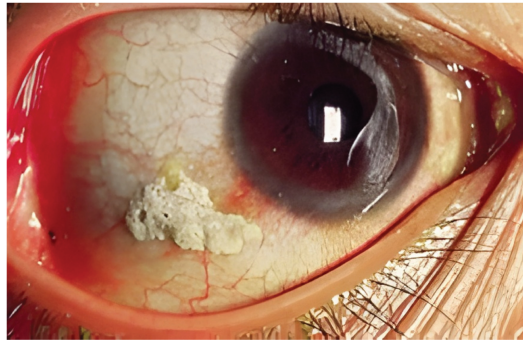
OK D. Vitamin K

WHY THIS IS RIGHT

Bulky, foul-smelling stools that stick to the pan indicate fat malabsorption (steatorrhea). Since vitamin K is a fat-soluble vitamin (A, D, E, K), its absorption is impaired in conditions causing fat malabsorption. Vitamin K is essential for γ -carboxylation of clotting factors II, VII, IX, and X, as well as proteins C and S. Deficiency leads to impaired coagulation and presents with bleeding tendencies. Additionally, some antidepressants (e.g., SSRIs) can increase bleeding risk by affecting platelet function, further worsening the bleeding in vitamin K deficiency.

QUESTION

| Condition shown below is due to which vitamin deficiency?



A Vitamin A

CORRECT

B Vitamin B

C Vitamin C

D Vitamin K

RIGHT ANSWER

OK A. Vitamin A

WHY THIS IS RIGHT

Image shows Bitot spots-foamy, whitish patches on the conjunctiva, which are characteristic of vitamin A deficiency and part of the spectrum of xerophthalmia.

- Vitamin A is essential for maintenance of epithelial integrity and mucin production in the conjunctiva.
- Deficiency leads to keratinization of conjunctival epithelium and accumulation of debris -> forming Bitot spots.
- It also impairs rhodopsin formation, causing night blindness.

Spectrum of Vitamin A deficiency:

1. Night blindness
2. Conjunctival xerosis
3. Bitot spots
4. Corneal xerosis
5. Keratomalacia (corneal softening -> blindness)

Why other options are incorrect:

- Vitamin B: Causes glossitis, neuropathy, anemia
- Vitamin C: Causes scurvy (bleeding gums, poor wound healing)
- Vitamin K: Causes bleeding due to clotting factor deficiency

QUESTION

FAD status in body is determined by?

- A** Thiamine
- B** **Glutathione reductase**
- C** Hexokinase
- D** Transketolase

CORRECT

RIGHT ANSWER

- OK** **B. Glutathione reductase**

WHY THIS IS RIGHT

FAD (Flavin adenine dinucleotide) is derived from riboflavin (vitamin B2) and functions as a cofactor in various redox reactions.

The functional status of FAD in the body is assessed by measuring the activity of glutathione reductase in red blood cells. This enzyme requires FAD as a cofactor to convert oxidized glutathione (GSSG) to reduced glutathione (GSH).

- In riboflavin (FAD) deficiency, glutathione reductase activity decreases.
- When FAD is added in vitro, enzyme activity increases (activation coefficient), indicating deficiency.

Why other options are incorrect:

- Thiamine: Assessed by transketolase activity (not FAD).
- Hexokinase: Involved in glycolysis, not used for vitamin assessment.
- Transketolase: Used to assess thiamine (vitamin B1) status.

QUESTION

A known patient of tuberculosis, on INH for the past 3 months complains of numbness, tingling and burning sensation in all the extremities. What could be vitamin deficiency?

A Vitamin B6

CORRECT

B Vitamin B12

C Vitamin B1

D Vitamin B3

RIGHT ANSWER

OK

A. Vitamin B6

WHY THIS IS RIGHT

Isoniazid (INH) therapy in tuberculosis can cause vitamin B6 (pyridoxine) deficiency, leading to peripheral neuropathy (numbness, tingling, burning sensation).

- INH forms complexes with pyridoxine and increases its excretion
- This leads to ↓ availability of pyridoxal phosphate (active B6)
- Pyridoxal phosphate is required for neurotransmitter synthesis (e.g., GABA)

Deficiency results in:

- Peripheral neuropathy
- Paresthesia (glove and stocking pattern)

Prevention: Always give prophylactic vitamin B6 (pyridoxine) with INH therapy

Why other options are incorrect:

- Vitamin B12: Causes neuropathy but not specifically linked to INH
- Vitamin B1 (Thiamine): Causes beriberi/Wernicke's
- Vitamin B3 (Niacin): Causes pellagra (dermatitis, diarrhea, dementia)

QUESTION

Identify the vitamin deficiency:



A Vitamin A

B Vitamin D

CORRECT

C Vitamin B3

D Vitamin B1

RIGHT ANSWER

OK B. Vitamin D

WHY THIS IS RIGHT

The image shows bowed legs (genu varum) in a child, which is a classical feature of rickets due to vitamin D deficiency.

- Vitamin D is essential for calcium and phosphate absorption from the intestine
- Deficiency -> ↓ calcium absorption -> defective mineralization of growing bones
- Leads to soft, weak bones that bend under body weight -> bowed legs

Other features of rickets:

- Rachitic rosary (beading at costochondral junctions)
- Widened wrists
- Frontal bossing
- Delayed growth

Why other options are incorrect:

- Vitamin A: Night blindness, xerophthalmia
- Vitamin B3 (Niacin): Pellagra (dermatitis, diarrhea, dementia)
- Vitamin B1 (Thiamine): Beriberi (neuropathy, cardiac issues)

QUESTION

A child presents with thinning of hair and scaling of the scalp. On taking further history, it is found that she consumes raw eggs in her diet regularly. The given manifestations are due to the deficiency of which of the following vitamins?

- A** Niacin
- B** Pyridoxine
- C** Biotin
- D** Thiamine

CORRECT

RIGHT ANSWER

OK C. Biotin

WHY THIS IS RIGHT

Consumption of raw eggs leads to biotin (vitamin B7) deficiency due to the presence of avidin, a protein in egg white that binds biotin and prevents its absorption.

Biotin functions as a coenzyme for carboxylation reactions:

- Pyruvate carboxylase
- Acetyl-CoA carboxylase
- Propionyl-CoA carboxylase

Clinical features of biotin deficiency:

- Alopecia (hair thinning/loss)
- Dermatitis (scaly skin, especially scalp)
- Neurological symptoms (depression, lethargy)

QUESTION

A patient presenting with bleeding gums and easy bruisability was diagnosed with scurvy. What is the biochemical basis of scurvy?

- A** Low calcium
- B** Defective collagen formation
- C** Inhibition of clotting factors
- D** Increased keratinization of epithelium

CORRECT

RIGHT ANSWER

- B. Defective collagen formation**

WHY THIS IS RIGHT

Scurvy is caused by vitamin C (ascorbic acid) deficiency, leading to defective collagen synthesis. Vitamin C is required for hydroxylation of proline and lysine residues in collagen via prolyl and lysyl hydroxylase. This hydroxylation is essential for:

- Stabilization of collagen triple helix
- Proper cross-linking of collagen fibers

Clinical features explained:

- Bleeding gums & petechiae -> weak vascular connective tissue
- Poor wound healing
- Bone pain (in children)

Why other options are incorrect:

- Low calcium: Causes bone disorders (rickets/osteomalacia)
- Inhibition of clotting factors: Seen in vitamin K deficiency
- Increased keratinization: Seen in vitamin A deficiency

QUESTION

A child was brought with pedal edema and cheilosis. Further investigations revealed cardiomegaly. What is the vitamin deficiency associated with this clinical presentation?

A Riboflavin

B Thiamine

CORRECT

C Pyridoxine

D Niacin

RIGHT ANSWER

OK

B. Thiamine

WHY THIS IS RIGHT

Thiamine (Vitamin B1) deficiency leads to beriberi, which can present with both cardiac and peripheral manifestations. Types relevant here:

- Wet beriberi:

- o Cardiomegaly, high-output cardiac failure
- o Edema (pedal edema)

- Dry beriberi:

- o Peripheral neuropathy

Thiamine is a cofactor for key enzymes in energy metabolism:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase

Cheilosis: Though classically seen in riboflavin deficiency, it can coexist in malnutrition, but the cardiomegaly + edema strongly points to thiamine deficiency.

Why other options are incorrect:

- Riboflavin: Causes cheilosis and glossitis but not cardiomegaly
- Pyridoxine: Causes neuropathy and sideroblastic anemia
- Niacin: Causes pellagra (dermatitis, diarrhea, dementia)

QUESTION

A chronic alcoholic patient presented to the casualty with confusion, ataxia, and painful eye movements. What could be the vitamin deficiency seen in this patient?

A Vitamin B deficiency

CORRECT

B Vitamin A deficiency

C Vitamin D deficiency

D Vitamin C deficiency

RIGHT ANSWER

OK A. Vitamin B deficiency

WHY THIS IS RIGHT

A chronic alcoholic presenting with confusion, ataxia, and painful/abnormal eye movements (ophthalmoplegia) is exhibiting the classic triad of Wernicke encephalopathy, caused by thiamine (Vitamin B1) deficiency.

Thiamine is a cofactor for key enzymes in glucose metabolism:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase

Deficiency $\rightarrow$ $\downarrow$ ATP production $\rightarrow$ neuronal energy failure, especially in brain regions like mammillary bodies and periaqueductal gray

Clinical triad:

- Confusion
- Ataxia
- Ophthalmoplegia (eye movement abnormalities)

Why other options are incorrect:

- Vitamin A: Vision problems (night blindness)
- Vitamin D: Bone disorders
- Vitamin C: Scurvy (bleeding gums)

QUESTION

A family consumes only polished rice. Which of the following combination of vitamin deficiency and enzymatic defect will be present in this family?

A Riboflavin - Glutathione Reductase

B Thiamine - Transketolase

CORRECT

C Thiamine - Transaminase

D Riboflavin - Transketolase

RIGHT ANSWER

B. Thiamine - Transketolase

WHY THIS IS RIGHT

A diet consisting predominantly of polished rice leads to thiamine (Vitamin B1) deficiency, because the outer bran layer (rich in thiamine) is removed during polishing.

Thiamine (as thiamine pyrophosphate, TPP) is a cofactor for several enzymes, including:

- Transketolase (in the HMP shunt)
- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase

Among these, transketolase activity in RBCs is commonly used to assess thiamine status.

Why other options are incorrect:

- Riboflavin - Glutathione reductase: Correct pair, but riboflavin deficiency is not the primary issue with polished rice diet
- Thiamine - Transaminase: Transaminases require vitamin B6 (pyridoxine)
- Riboflavin - Transketolase: Transketolase is dependent on thiamine, not riboflavin

QUESTION

A patient presented with diarrhea, dermatitis, and dementia. Which of the following vitamin deficiencies would you suspect in this patient?

A Vitamin B3 deficiency

CORRECT

B Vitamin B1 deficiency

C Vitamin B6 deficiency

D Vitamin B12 deficiency

RIGHT ANSWER

OK A. Vitamin B3 deficiency

WHY THIS IS RIGHT

The triad of diarrhea, dermatitis, and dementia is characteristic of pellagra, caused by niacin (Vitamin B3) deficiency.

- Precursor of NAD⁺ and NADP⁺
- Essential for redox reactions and energy metabolism
- Affects tissues with high turnover -> skin, GI tract, and brain

Clinical features (3 D's):

- Dermatitis: Photosensitive, hyperpigmented rash (Casal necklace)
- Diarrhea: Due to GI mucosal atrophy
- Dementia: Neurological impairment

(+ 4th D: Death if untreated)

Why other options are incorrect:

- Vitamin B1: Beriberi, Wernicke's encephalopathy
- Vitamin B6: Neuropathy, sideroblastic anemia
- Vitamin B12: Megaloblastic anemia + neurological deficits (no dermatitis/diarrhea triad)

QUESTION

Which of the following zymogens requires gamma-glutamyl carboxylase?

- A** Factors II, VIII, IX, X
- B** Factors II, VII, IX, and X CORRECT
- C** Factors II, VII, IX, XI
- D** Factors II, VIII, X, XI

RIGHT ANSWER

- B. Factors II, VII, IX, and X**

WHY THIS IS RIGHT

γ -Glutamyl carboxylase is a vitamin K-dependent enzyme that catalyzes the γ -carboxylation of glutamate residues on certain clotting factors. This modification allows these factors to bind calcium (Ca^{2+}) and attach to phospholipid surfaces, which is essential for normal coagulation cascade function. Vitamin K-dependent clotting factors:

- Factor II (Prothrombin)
- Factor VII
- Factor IX
- Factor X

(+ Proteins C and S)

Clinical relevance:

- Vitamin K deficiency or warfarin therapy $\rightarrow$ impaired γ -carboxylation $\rightarrow$ bleeding tendency
- First factor to be affected: Factor VII (shortest half-life $\rightarrow$ $\uparrow$ PT earliest)

Why other options are incorrect:

- Factor VIII: Not vitamin K dependent
- Factor XI: Not vitamin K dependent

QUESTION

A 12-year-old adolescent presents with progressive joint pain, easy fatigability, and weakness. On examination, the patient has marfanoid habitus and mild developmental delay. Laboratory evaluation shows elevated homocysteine levels with decreased cysteine levels. Which vitamin supplementation is most appropriate in this condition?

A Vitamin B6 (Pyridoxine)

CORRECT

B Vitamin B2 (Riboflavin)

C Vitamin B1 (Thiamine)

D Vitamin B7 (Biotin)

RIGHT ANSWER

OK **A. Vitamin B6 (Pyridoxine)**

WHY THIS IS RIGHT

In homocystinuria, elevated homocysteine with low cysteine indicates a defect in the transsulfuration pathway, specifically the enzyme cystathionine β -synthase (CBS).

- Vitamin B6 (PLP) is a cofactor for CBS
- Required for conversion of:

Homocysteine $\rightarrow$ Cystathionine $\rightarrow$ Cysteine

- CBS deficiency $\rightarrow$ $\uparrow$ homocysteine and $\downarrow$ cysteine
- Leads to vascular damage, thrombosis, skeletal abnormalities

Giving Vitamin B6 enhances residual enzyme activity, reducing homocysteine levels

Clinical features of homocystinuria:

- Marfanoid habitus
- Lens subluxation (downward)
- Thromboembolism
- Intellectual disability

Why other options are incorrect:

- Vitamin B2: FAD-related enzymes
- Vitamin B1: Carbohydrate metabolism
- Vitamin B7 (Biotin): Carboxylation reactions

QUESTION

| Which vitamin can be deficient in a patient who has undergone gastrectomy?

A Vitamin A

B **Vitamin B12**

CORRECT

C Vitamin C

D Vitamin D

RIGHT ANSWER

OK **B. Vitamin B12**

WHY THIS IS RIGHT

Vitamin B12 (cobalamin) deficiency is common after gastrectomy due to loss of intrinsic factor (IF) production.

- Parietal cells of stomach produce intrinsic factor
- IF is essential for absorption of vitamin B12 in the terminal ileum
- Gastrectomy -> ↓ intrinsic factor -> impaired B12 absorption

Consequences:

- Megaloblastic anemia
- Neurological manifestations (subacute combined degeneration)
 - o Paresthesia
 - o Loss of vibration & position sense
 - o UMN signs

Why other options are incorrect:

- Vitamin A: Fat-soluble, absorbed in intestine
- Vitamin C: Absorbed directly, not IF-dependent
- Vitamin D: Fat-soluble, not affected by intrinsic factor

QUESTION

A 45-year-old woman with Rheumatoid arthritis is started on low-dose methotrexate therapy. After a few weeks, she develops oral ulcers and mild bone marrow suppression. Which of the following is most directly reduced due to this mechanism?

A Thymidine monophosphate (TMP)

CORRECT

B Guanosine monophosphate (GMP)

C Cytidine monophosphate (CMP)

D Adenosine monophosphate (AMP)

RIGHT ANSWER

OK

A. Thymidine monophosphate (TMP)

WHY THIS IS RIGHT

Methotrexate is a folate antagonist that inhibits the enzyme dihydrofolate reductase (DHFR).

- Blocks conversion of dihydrofolate (DHF) → tetrahydrofolate (THF)
- THF is required for one-carbon transfer reactions
- Impaired synthesis of thymidylate (dTMP) from dUMP
- Leads to ↓ DNA synthesis, especially in rapidly dividing cells

Clinical relevance:

- Used in cancer chemotherapy, autoimmune diseases (RA)
- Causes megaloblastic anemia as a side effect

Why other options are incorrect:

- GMP, AMP: Purine synthesis also affected indirectly but main tested effect is dTMP inhibition
- CMP: Pyrimidine synthesis not directly dependent on folate in this step

QUESTION

A patient presented with follicular hyperkeratosis on the extensor aspect of the forearm. What is the diagnosis?

A Darier's disease

B Phrynoderma

CORRECT

C Eruptive xanthoma

D Folliculitis

RIGHT ANSWER

B. Phrynoderma

WHY THIS IS RIGHT

Phrynoderma ("toad skin") is characterized by follicular hyperkeratosis on extensor surfaces (arms, thighs), most commonly due to vitamin A deficiency.

- Vitamin A is essential for normal epithelial differentiation
- Deficiency -> abnormal keratinization of hair follicles -> rough, dry papules

Clinical features:

- Rough, keratotic papules over extensor surfaces
- Dry, coarse skin
- May be associated with night blindness

Why other options are incorrect:

- Darier's disease: Genetic disorder with greasy papules (seborrheic areas)
- Eruptive xanthoma: Yellow papules due to hypertriglyceridemia
- Folliculitis: Infection of hair follicles (pus-filled lesions)

QUESTION

In the shown image, which one of the following is a source of vitamin B1?



A A

CORRECT

B B

C C

D D

RIGHT ANSWER

OK A. A

WHY THIS IS RIGHT

Vitamin B1 (Thiamine) is primarily obtained from whole grains, cereals, and pulses (unpolished rice, legumes)-represented by option A in the image.

Thiamine (as thiamine pyrophosphate, TPP) acts as a cofactor for:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase (HMP shunt)

Deficiency leads to:

- Beriberi (dry: neuropathy, wet: cardiac failure)
- Wernicke-Korsakoff syndrome (in alcoholics)

QUESTION

The treatment given to the patient presenting with a history of binge alcohol intake and symptoms of convulsions, altered sensorium, and plasma glucose level of 45 mg/dL is:

A 25% dextrose

B Fomepizole

C Thiamine

D Thiamine followed by dextrose

CORRECT

RIGHT ANSWER

OK

D. Thiamine followed by dextrose

WHY THIS IS RIGHT

In a chronic alcoholic with hypoglycemia (glucose 45 mg/dL) and altered sensorium, the correct management is thiamine administration before glucose.

- Chronic alcoholics are commonly thiamine (Vitamin B1) deficient
- Thiamine is required for glucose metabolism (pyruvate dehydrogenase, etc.)
- Giving glucose first increases metabolic demand and can precipitate or worsen Wernicke encephalopathy

Correct sequence: Thiamine first -> then dextrose

- Thiamine deficiency -> impaired cerebral energy metabolism
- Glucose without thiamine -> accumulation of lactate -> neuronal injury

Why other options are incorrect:

- 25% dextrose alone: Can worsen neurological damage
- Fomepizole: Used in methanol/ethylene glycol poisoning
- Thiamine alone: Does not correct hypoglycemia

QUESTION

Which micronutrient deficiency causes anemia, bleeding gums, and white line of Frenkel on X-ray?

A Copper

B Vitamin C

CORRECT

C Iron

D Zinc

RIGHT ANSWER

OK B. Vitamin C

WHY THIS IS RIGHT

The triad of anemia, bleeding gums, and “white line of Frankel” on X-ray is characteristic of vitamin C (ascorbic acid) deficiency -> scurvy.

Vitamin C is essential for hydroxylation of proline and lysine in collagen via:

- Prolyl hydroxylase
- Lysyl hydroxylase

Deficiency -> defective collagen synthesis -> weak connective tissue.

Clinical features explained:

- Bleeding gums & petechiae: Fragile capillaries due to weak collagen
- Anemia: Due to chronic bleeding + ↓ iron absorption (vitamin C enhances iron absorption)
- White line of Frankel (dense metaphyseal line): Due to defective osteoid formation in growing bones

Radiological signs in scurvy:

- White line of Frankel
- Pelkan spurs
- Scorbutic rosary

Why other options are incorrect:

- Copper: Causes anemia and neurological issues, not bleeding gums + Frankel line
- Iron: Causes anemia only
- Zinc: Causes dermatitis and poor wound healing

QUESTION

A patient present with symptoms of dermatitis, diarrhea, and cognitive decline. Which micronutrient deficiency is most likely responsible?



A Niacin

CORRECT

B Thiamine

C Pyridoxine

D Retinol

RIGHT ANSWER

OK A. Niacin

WHY THIS IS RIGHT

The image shows Casal necklace—a photosensitive hyperpigmented dermatitis around the neck, which is characteristic of pellagra due to niacin (Vitamin B3) deficiency.

Niacin is a precursor of NAD⁺ and NADP⁺, essential for oxidation-reduction reactions and energy metabolism.

Deficiency → impaired cellular energy production, especially in skin, GI tract, and brain.

Classic triad (3 D's):

- Dermatitis (photosensitive, Casal necklace)
- Diarrhea
- Dementia

(+ Death if untreated)

Why other options are incorrect:

- Thiamine: Beriberi, Wernicke's encephalopathy
- Pyridoxine: Neuropathy, sideroblastic anemia
- Retinol (Vitamin A): Night blindness, xerophthalmia

QUESTION

Which amino acid will not be synthesized due to B12 deficiency in a patient with history of ileectomy with elevated homocysteine levels, increased methylmalonyl CoA and peripheral neuropathy?

A Methionine

CORRECT

B Cysteine

C Glutathione

D Tryptophan

RIGHT ANSWER

OK **A. Methionine**

WHY THIS IS RIGHT

Vitamin B12 deficiency (e.g., after ileectomy, where B12 is absorbed) leads to impairment of two key reactions:

1. Homocysteine → Methionine (via methionine synthase, B12-dependent)
2. Methylmalonyl-CoA → Succinyl-CoA (via methylmalonyl-CoA mutase)

Biochemical consequences:

- ↑ Homocysteine (cannot convert to methionine)
- ↓ Methionine synthesis
- ↑ Methylmalonyl-CoA → contributes to neurological damage

Clinical relevance:

- Peripheral neuropathy (due to myelin damage)
- Megaloblastic anemia
- Seen in ileal resection (ileectomy) due to impaired B12 absorption

Why other options are incorrect:

- Cysteine: Formed from homocysteine (via B6-dependent pathway), not directly dependent on B12
- Glutathione: Synthesized from glutamate, cysteine, glycine
- Tryptophan: Essential amino acid (not synthesized in body)

QUESTION

A non-alcoholic hypertensive patient on hydrochlorothiazide therapy presents with cerebellar ataxia, peripheral neuropathy, symmetrical tingling sensations, cardiac failure, and pedal edema. Deficiency of which of the following vitamins or nutrients can cause similar clinical features?

A Selenium

B Thiamine

CORRECT

C Vit B12

D Vit B2

RIGHT ANSWER

OK B. Thiamine

WHY THIS IS RIGHT

The constellation of cerebellar ataxia, peripheral neuropathy, tingling, cardiac failure, and pedal edema is characteristic of thiamine (Vitamin B1) deficiency, which presents as a combination of dry beriberi (neuropathy) and wet beriberi (cardiac failure + edema).

Chronic diuretic use can increase urinary loss of thiamine, predisposing to deficiency

Thiamine (as TPP) is a cofactor for:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase

Deficiency -> impaired ATP production, especially affecting:

- Nervous system -> neuropathy, ataxia
- Heart -> cardiomyopathy, heart failure, edema

Why other options are incorrect:

- Selenium: Cardiomyopathy (Keshan disease) but no neuropathy triad
- Vitamin B12: Neuropathy without cardiac failure/edema
- Vitamin B2: Cheilosis, glossitis, dermatitis

QUESTION

A patient presents with fatigue, gum bleeding, and multiple perifollicular hemorrhages on the lower limbs, along with a bleeding tendency. These findings are most consistent with a deficiency of which enzyme activity?

A Prolyl hydroxylase

CORRECT

B Lysyl oxidase

C Tyrosinase

D Alkaline phosphatase

RIGHT ANSWER

OK **A. Prolyl hydroxylase**

WHY THIS IS RIGHT

The clinical features-bleeding gums, perifollicular hemorrhages, easy bruising, and fatigue-are classic for scurvy (vitamin C deficiency).

Vitamin C is required as a cofactor for:

- Prolyl hydroxylase
- Lysyl hydroxylase

These enzymes catalyze hydroxylation of proline and lysine residues in collagen, which is essential for:

- Stability of the collagen triple helix
- Proper cross-linking of collagen fibers

Why other options are incorrect:

- Lysyl oxidase: Involved in collagen cross-linking but is copper-dependent, not vitamin C dependent
- Tyrosinase: Involved in melanin synthesis
- Alkaline phosphatase: Associated with bone metabolism

QUESTION

| Which of the following statements regarding folate deficiency is FALSE?

- A** High homocysteine and normal methyl malonic acid
- B** If given at the diagnosis of pregnancy - can prevent neural tube defects CORRECT
- C** Folate supplementation is required in children with chronic hemolytic anemia
- D** Vitamin B12 deficiency should be corrected before supplementing folate

RIGHT ANSWER

- OK** **B. If given at the diagnosis of pregnancy - can prevent neural tube defects**

WHY THIS IS RIGHT

Folate is essential for DNA synthesis and neural tube development. To prevent neural tube defects (NTDs) such as spina bifida and anencephaly, folic acid supplementation must begin before conception and during early pregnancy (first 4 weeks), when neural tube closure occurs. Therefore, starting folate only at the time of pregnancy diagnosis may be too late to prevent NTDs, making that statement false.

Other statements are true:

- Folate deficiency: ↑ homocysteine with normal methylmalonic acid.
- Chronic hemolytic anemia: Increased RBC turnover -> higher folate requirement.
- Vitamin B12 deficiency should be corrected before folate therapy to avoid masking B12 deficiency and worsening neurological damage.

QUESTION

Korsakoff psychosis is true except?

1. Amnesia
2. Confabulation
3. Ophthalmoplegia
4. Ataxia

A 1, 2, 3

B 2, 3, 4

C 1, 3

D 3, 4

CORRECT

RIGHT ANSWER

OK D. 3, 4

WHY THIS IS RIGHT

Korsakoff psychosis is a chronic neuropsychiatric disorder caused by thiamine (vitamin B₁) deficiency, most commonly seen in chronic alcoholism. It is characterized by anterograde and retrograde amnesia with confabulation (fabrication of stories to fill memory gaps). Ophthalmoplegia and ataxia are features of Wernicke encephalopathy, the acute stage of thiamine deficiency.

QUESTION

In folate deficiency, the False statement is:

- A** High homocysteine and normal methylmalonic acid
- B** Hemolytic anemia is a feature CORRECT
- C** B12 supplementation is recommended along with Folate
- D** Purine and pyrimidine synthesis are affected

RIGHT ANSWER

- OK** **B. Hemolytic anemia is a feature**

WHY THIS IS RIGHT

Folate deficiency causes impaired DNA synthesis due to defective purine and thymidylate (pyrimidine) synthesis, leading to megaloblastic anemia. It is characterized by elevated homocysteine levels with normal methylmalonic acid (unlike vitamin B₁₂ deficiency).

Hemolytic anemia is not a typical feature; instead, folate deficiency produces megaloblastic anemia due to ineffective erythropoiesis. Vitamin B₁₂ is often supplemented with folate to avoid masking an underlying B₁₂ deficiency and preventing neurological complications.

QUESTION

A 52-year-old strict vegetarian presents with progressive numbness in feet, gait instability, and memory changes. Examination reveals loss of vibration and position sense with spasticity in lower limbs. Blood tests show macrocytic anemia. He is started on folic acid therapy, after which his anemia improves, but neurological symptoms worsen.

What is the most likely mechanism for this deterioration?

A Folate trap leading to methyl-THF accumulation

CORRECT

B Depletion of S-adenosylmethionine (SAM)

C Pernicious anemia

D Homocysteine accumulation

RIGHT ANSWER

OK A. Folate trap leading to methyl-THF accumulation

WHY THIS IS RIGHT

In vitamin B₁₂ deficiency, folate becomes trapped as methyl-tetrahydrofolate (methyl-THF) due to impaired conversion of homocysteine to methionine, a reaction that requires vitamin B₁₂.

- Methionine synthase requires vitamin B₁₂
- Without B₁₂ -> methyl-THF cannot convert to THF
- Leads to "folate trap" with accumulation of methyl-THF

Clinical Significance:

- Folate supplementation corrects anemia by providing folate for DNA synthesis
- But it does not correct B₁₂ deficiency
- Neurologic damage (due to impaired myelin synthesis) may worsen or continue

QUESTION

Vitamin leading to diversion of 3-hydroxykynurenine toward xanthurenic acid:

CORRECT

A Pyridoxine

B Pantothenic acid

C Thiamine

D Biotin

RIGHT ANSWER

A. Pyridoxine

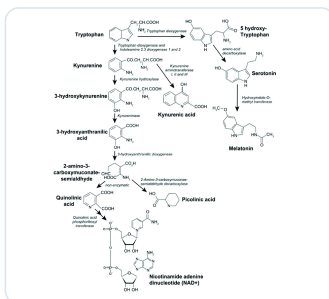
WHY THIS IS RIGHT

Pyridoxine (Vitamin B6) is required as a cofactor in the tryptophan-kynurenine pathway. It is necessary for the enzyme kynureninase, which converts 3-hydroxykynurenine -> 3-hydroxyanthranilic acid.

When pyridoxine is deficient:

- The normal pathway is blocked.
- 3-hydroxykynurenine is diverted to form xanthurenic acid.
- Increased urinary excretion of xanthurenic acid occurs.

Can occur with isoniazid therapy, which interferes with pyridoxine metabolism.



QUESTION

A 35-year-old female is hospitalized with headaches and vomiting. She has a long history of psychiatric illness and is known to practice eccentric dietary habits. Physical findings include papilledema, dry skin, and hepatosplenomegaly. Head CT scan is ordered immediately but is negative for intracranial mass. Which of the following is a likely cause of this patient's condition?

A Thiamine deficiency

B Riboflavin deficiency

C Vitamin C overuse

D Vitamin A overuse

CORRECT

RIGHT ANSWER

OK D. Vitamin A overuse

WHY THIS IS RIGHT

This patient has hypervitaminosis A, suggested by headaches, vomiting, papilledema (pseudotumor cerebri), hepatosplenomegaly, dry skin, and psychiatric issues with eccentric diet. Excess vitamin A leads to increased intracranial pressure and systemic toxicity affecting the skin and liver. Head CT is normal, which is typical for pseudotumor cerebri caused by vitamin A toxicity.

Why the others are incorrect:

- A. Thiamine deficiency: Causes Wernicke-Korsakoff syndrome or beriberi; does not produce papilledema or hepatosplenomegaly.
- B. Riboflavin deficiency: Leads to cheilitis, glossitis, dermatitis, not intracranial hypertension.
- C. Vitamin C overuse: Can cause kidney stones or GI upset, not pseudotumor cerebri.

QUESTION

31-year-old previously healthy man comes to the OPD due to myalgias, anorexia, and skin rash. The patient works as a personal trainer and is a bodybuilding enthusiast. He denies using anabolic steroids but has been consuming large amounts of raw egg whites for the past several months. Physical examination shows macular dermatitis of the extremities. A water-soluble vitamin deficiency is suspected as the cause of his condition. Which of the following biochemical conversions most likely uses the deficient vitamin as a cofactor?

A Glucose to ribose-5-phosphate

B Pyruvate to acetyl-CoA

C Pyruvate to alanine

D Pyruvate to oxaloacetate

CORRECT

RIGHT ANSWER

OK

D. Pyruvate to oxaloacetate

WHY THIS IS RIGHT

This patient has biotin deficiency, suggested by chronic consumption of raw egg whites (which contain avidin that binds biotin), along with dermatitis, myalgias, and anorexia. Biotin acts as a cofactor for carboxylase enzymes, including pyruvate carboxylase, which converts pyruvate to oxaloacetate in gluconeogenesis. This reaction is critical for maintaining glucose production and TCA cycle intermediates.

Why the others are incorrect:

A. Glucose → ribose-5-phosphate: Requires transketolase (thiamine), not biotin.

B. Pyruvate → acetyl-CoA: Catalyzed by pyruvate dehydrogenase, which uses thiamine, lipoate, FAD, and NAD⁺.

C. Pyruvate → alanine: Catalyzed by alanine aminotransferase, requiring pyridoxal phosphate (B6), not biotin.

QUESTION

A research scientist is studying biochemical reactions that take place in the liver. He cultures hepatocytes in a growth media enriched with glutamate labelled with nitrogen isotopes. After some time, he finds that the nitrogen isotopes are transferred to oxaloacetate, forming aspartate in the process. Which of the following substances is most likely involved in this reaction?

A Biotin

B Folic acid

C Niacin

D Pyridoxine

CORRECT

RIGHT ANSWER

OK

D. Pyridoxine

WHY THIS IS RIGHT

The reaction described is a transamination, where an amino group from glutamate is transferred to oxaloacetate to form aspartate. This process is catalyzed by aminotransferases, which require pyridoxal phosphate (the active form of pyridoxine) as a cofactor. Pyridoxine is essential for all amino acid metabolism involving transamination.

Why the others are incorrect:

A. Biotin: Cofactor for carboxylation reactions (e.g., pyruvate → oxaloacetate), not transamination.

B. Folic acid: Involved in one-carbon transfers, nucleotide synthesis, and methylation reactions; not directly in amino group transfer.

C. Niacin: Precursor for NAD⁺/NADP⁺, used in redox reactions, not in nitrogen transfer.

QUESTION

A 5-year-old boy with developmental delay is brought to the OPD due to difficulty "seeing the board" at school. Examination shows a tall, thin habitus with elongated limbs. Funduscopy shows bilateral lens subluxation. Four years later, the patient dies suddenly of a massive cerebrovascular accident. Autopsy shows middle cerebral artery thrombosis and old renal infarcts. His parents wish to know if anything could have been done to have prevented his death. Which of the following would have been the most appropriate supplementation for this patient?

A Ascorbic acid

B Carnitine

C Pyridoxine

CORRECT

D Thiamine

RIGHT ANSWER

OK C. Pyridoxine

WHY THIS IS RIGHT

This child has homocystinuria, suggested by tall stature, lens subluxation, developmental delay, and premature thromboembolic events. The most common form is due to cystathionine β -synthase deficiency, which leads to elevated homocysteine and methionine. Supplementation with pyridoxine, a cofactor for CBS, can lower homocysteine levels in responsive patients and reduce the risk of thrombosis, potentially preventing early death.

Why the others are incorrect:

- A. Ascorbic acid: Used for collagen hydroxylation defects (e.g., scurvy), not homocystinuria.
- B. Carnitine: Helps with fatty acid metabolism; not effective in CBS deficiency.
- D. Thiamine: Coenzyme for pyruvate dehydrogenase; unrelated to homocysteine metabolism.

QUESTION

The sequence of vitamin D synthesis is:

A Liver - Skin - Kidney

B Skin - Liver - Kidney

CORRECT

C Skin - Kidney - Liver

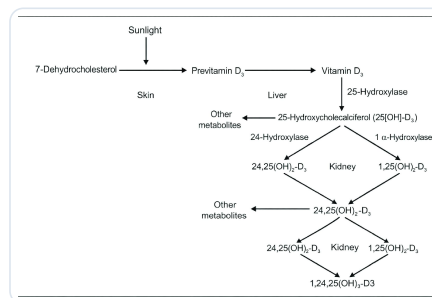
D Liver - Kidney - Skin

RIGHT ANSWER

OK B. Skin - Liver - Kidney

WHY THIS IS RIGHT

The synthesis of vitamin D follows the sequence of Skin -> Liver -> Kidney, with each organ playing a critical role in converting the precursor to its active form.



QUESTION

A child presents with thinning of hair and scaling of the scalp. On taking further history, it is found that she consumes raw eggs in her diet regularly. The given manifestations are due to the defect in which of the following reactions?

A Pyruvate carboxylase

CORRECT

B Pyruvate dehydrogenase

C Glycogen phosphorylase

D Glutathione reductase

RIGHT ANSWER

OK

A. Pyruvate carboxylase

WHY THIS IS RIGHT

This child is likely suffering from biotin deficiency due to avidin in raw egg whites binding biotin. Biotin is a cofactor for carboxylase enzymes, including pyruvate carboxylase, which catalyzes pyruvate $\rightarrow$ oxaloacetate in gluconeogenesis. Deficiency leads to impaired gluconeogenesis, metabolic acidosis, thinning hair, and dermatitis (scaling of scalp).

Why other options are incorrect:

B. Pyruvate dehydrogenase: Thiamine and lipoic acid-dependent; not affected by biotin.

C. Glycogen phosphorylase: Vitamin B6-dependent; unrelated to biotin deficiency.

D. Glutathione reductase: FAD-dependent; not affected by biotin deficiency.

QUESTION

A patient with celiac disease presents with peripheral neuropathy and decreased vibration and position sense. Investigations show hemolytic anemia and normal methylmalonic acid levels. Which of the following is most likely deficient in this patient?

A Pyridoxine

B Folate

C Vitamin B12

D Vitamin E

CORRECT

RIGHT ANSWER

OK

D. Vitamin E

WHY THIS IS RIGHT

This patient has peripheral neuropathy with loss of vibration and position sense, along with hemolytic anemia but normal methylmalonic acid, which points toward vitamin E deficiency. Vitamin E (α -tocopherol) is a lipid-soluble antioxidant, and its deficiency—commonly seen in fat malabsorption disorders like celiac disease—leads to:

- Hemolytic anemia due to oxidative damage to RBC membranes.
- Neurologic symptoms (spinocerebellar and posterior column dysfunction) causing loss of vibration and proprioception.

Why other options are incorrect:

A. Pyridoxine (B₆): Causes neuropathy but usually without hemolytic anemia.

B. Folate: Causes megaloblastic anemia, not hemolytic anemia or posterior column neuropathy.

C. Vitamin B₁₂: Deficiency elevates MMA, which is normal here, ruling it out.

QUESTION

A 45-year-old homeless man comes to the emergency department due to a "pins-and-needles" sensation in his legs. He also has painful lesions on his lips and corners of his mouth. Abdominal examination reveals hepatomegaly. Laboratory evaluation shows very low urinary riboflavin excretion. Activity of which of the following enzymes is most likely directly impaired in this patient?

A Glucose-6-phosphate dehydrogenase

B HMG-CoA reductase

C Isocitrate dehydrogenase

D Succinate dehydrogenase

CORRECT

RIGHT ANSWER

OK **D. Succinate dehydrogenase**

WHY THIS IS RIGHT

Riboflavin (vitamin B₂) is the precursor for FAD and FMN, which are essential cofactors for flavoproteins. Succinate dehydrogenase (Complex II of the ETC) is an FAD-dependent enzyme, and its activity is directly impaired in riboflavin deficiency. This leads to defects in oxidative phosphorylation and energy metabolism, which contributes to neuropathy and muscle weakness seen in this patient.

Why other options are incorrect:

- A. Glucose-6-phosphate dehydrogenase: NADP⁺-dependent; indirectly affected by riboflavin deficiency but not directly FAD-dependent.
- B. HMG-CoA reductase: Requires NADPH, not FAD/FMN.
- C. Isocitrate dehydrogenase: Uses NAD⁺ or NADP⁺, not FAD/FMN.

QUESTION

A 32-year-old previously healthy man comes to the emergency department due to a 3-hour history of chest pain, diaphoresis, and dyspnea. He does not smoke, exercises regularly, and eats a balanced diet. His father died at age 56 from a myocardial infarction. His blood pressure is 110/70 mm Hg and pulse is 110/min and regular. ECG shows ST elevation in the anterolateral leads. Coronary angiogram reveals proximal left anterior descending artery stenosis and thrombosis, which is treated with angioplasty and stent placement. Laboratory results are as follows:

Total cholesterol: 160 mg/dL

Low-density lipoprotein: 90 mg/dL

Glucose, serum: 98 mg/dL

Homocysteine, plasma: 21.5 $\mu\text{mol/L}$ (normal: 4-14 $\mu\text{mol/L}$)

Further testing reveals a homozygous mutation in the methylene tetrahydrofolate reductase gene that leads to decreased enzymatic activity. Due to this defect, the patient most likely has impairment converting homocysteine to which of the following?

A Cystathionine

B Cysteine

C Methionine

CORRECT

D Methylmalonyl-CoA

RIGHT ANSWER

OK C. Methionine

WHY THIS IS RIGHT

This patient has elevated homocysteine and a homozygous MTHFR mutation, which reduces activity of methylene tetrahydrofolate reductase. MTHFR generates 5-methyltetrahydrofolate, the methyl donor required by methionine synthase to convert homocysteine $\rightarrow$ methionine. Deficiency leads to hyperhomocysteinemia, a risk factor for premature atherosclerosis and thrombosis, as seen in this patient with early-onset MI.

Why other options are incorrect:

A. Cystathionine: Formed from homocysteine via cystathionine β -synthase, which requires vitamin B₁₂; not directly affected by MTHFR.

B. Cysteine: Downstream product of the transsulfuration pathway; secondary to cystathionine.

D. Methylmalonyl-CoA: Formed from propionyl-CoA; requires vitamin B₁₂ but unrelated to homocysteine metabolism.

QUESTION

Which of the following are vitamin B12-dependent enzymes?

1. Methyl malonyl CoA mutase
2. Homocysteine synthase
3. Propionyl CoA carboxylase
4. Methionine synthase

A 1, 2, 3 and 4

B 1 and 4 only

CORRECT

C 1 and 3 only

D 1, 2, and 3

RIGHT ANSWER

OK **B. 1 and 4 only**

WHY THIS IS RIGHT

Vitamin B₁₂ (cobalamin) is required as a cofactor for two key enzymes in human metabolism. Methylmalonyl-CoA mutase converts methylmalonyl-CoA to succinyl-CoA in odd-chain fatty acid and amino acid metabolism. Methionine synthase catalyzes the remethylation of homocysteine to methionine, linking B₁₂ deficiency to hyperhomocysteinemia and megaloblastic anemia.

Why other options are incorrect:

- 2\ Homocysteine synthase: Not a human enzyme; homocysteine is remethylated by methionine synthase.
- 3\ Propionyl-CoA carboxylase: Biotin-dependent, not B₁₂-dependent.

QUESTION

Which of the following is the earliest sign of riboflavin deficiency?

- A** Cheilosis
- B** Glossitis
- C** Seborrheic dermatitis
- D** Proliferation of the bulbar conjunctival capillaries

CORRECT

RIGHT ANSWER

- OK** **D. Proliferation of the bulbar conjunctival capillaries**

WHY THIS IS RIGHT

Riboflavin (vitamin B₂) deficiency initially affects rapidly dividing epithelial tissues and the eyes. The earliest clinical sign is dilatation and proliferation of bulbar conjunctival capillaries, giving a bloodshot appearance. This occurs before more obvious mucocutaneous manifestations and reflects impaired flavoprotein-dependent oxidative reactions.

Why other options are incorrect:

- A. Cheilosis: Occurs later as part of angular stomatitis.
- B. Glossitis: A later mucosal manifestation of riboflavin deficiency.
- C. Seborrheic dermatitis: Develops in more advanced deficiency, not an early sign.

QUESTION

Which of the following metabolic processes is not directly supported by vitamin B6 in its active form, pyridoxal 5'-phosphate (PLP)?

- A** Transamination of amino acids
- B** Decarboxylation of amino acids
- C** Breakdown of glycogen to glucose-1-phosphate

D Conversion of glucose to glycogen

CORRECT

RIGHT ANSWER

OK D. Conversion of glucose to glycogen

WHY THIS IS RIGHT

- Vitamin B6 in its active form, pyridoxal 5'-phosphate (PLP), serves as a coenzyme in multiple amino acid metabolism reactions and in glycogenolysis.
- PLP is essential for transamination and decarboxylation reactions, facilitating amino acid interconversion and neurotransmitter synthesis. It also acts as a cofactor for glycogen phosphorylase, which breaks glycogen into glucose-1-phosphate. However, glycogen synthesis (glycogenesis)-the conversion of glucose to glycogen-does not require PLP and instead involves enzymes like glycogen synthase and UDP-glucose pyrophosphorylase.

Why other options are incorrect:

- A. Transamination: PLP-dependent.
- B. Decarboxylation: PLP-dependent.
- C. Glycogen breakdown: Requires PLP via glycogen phosphorylase.

QUESTION

A 28-year-old man presents with fatigue, pallor, and ringed sideroblasts on bone marrow biopsy. Lab findings show increased serum iron and decreased hemoglobin. Which statement correctly differentiates ALA synthase I (ALAS-1) and ALA synthase II (ALAS-2) regulation in this patient?

A ALAS-1 is expressed in erythroid cells and activated by iron availability.

B ALAS-2 is expressed in the liver and inhibited by heme accumulation.

C ALAS-2 is erythroid-specific and requires pyridoxal phosphate.

CORRECT

D ALAS-1 is regulated by iron through iron-responsive elements.

RIGHT ANSWER

OK C. ALAS-2 is erythroid-specific and requires pyridoxal phosphate.

WHY THIS IS RIGHT

Aminolevulinic acid (ALA) synthase

ALA synthase is a mitochondrial enzyme. Two isoforms:

ALA-S1

- Located mostly in liver cells and to a smaller extent in other cells in the body
- Heme production for the liver cytochrome P450 mono-oxygenase system
- Gene is located on chromosome-3
- Loss-of-function mutation in the ALA-S1 gene causes impaired metabolism of xenobiotics

ALA-S2

- Produced in erythroid tissue
- Heme production for RBC hemoglobin
- Gene is located on the X-chromosome
- Loss-of-function mutations in ALA-S2 result in anemia

Porphyrin biosynthesis is primarily regulated via ALA-S regulation.

QUESTION

| Which type of PCR is suitable for identifying multiple causative agents?

A Conventional PCR

B Real-time PCR

C **Multiplex PCR**

CORRECT

D Nested PCR

RIGHT ANSWER

OK **C. Multiplex PCR**

WHY THIS IS RIGHT

Multiplex PCR is used for simultaneous detection of multiple pathogens in a single reaction by using multiple sets of primers targeting different DNA sequences.

- Principle:

- o Multiple primer pairs -> amplify different targets in one tube
- o Allows identification of multiple causative agents at once

- Advantages:

- o Saves time and cost
- o Requires smaller sample volume
- o Useful in syndromic diagnosis (e.g., respiratory, GI infections)

- Why not others:

- o Conventional PCR: detects single target per reaction
- o Real-time PCR: quantitative, but usually single target (unless multiplexed)
- o Nested PCR: increases sensitivity/specificity, not for multiple agents

QUESTION

A patient presents with multiple colonic polyps and is diagnosed with colorectal carcinoma. There is a strong family history suggestive of Hereditary Non-Polyposis Colorectal Cancer (HNPCC). Which DNA repair mechanism is most likely defective in this condition?

A Nucleotide excision repair

B Mismatch repair

CORRECT

C Base excision repair

D Double-strand break repair

RIGHT ANSWER

B. Mismatch repair

WHY THIS IS RIGHT

Hereditary Non-Polyposis Colorectal Cancer (HNPCC), also called Lynch syndrome, is caused by defects in the DNA mismatch repair (MMR) system. This repair mechanism normally corrects base-base mismatches and small insertion/deletion errors that occur during DNA replication.

Common genes involved:

- MLH1
- MSH2
- MSH6
- PMS2

Why other options are incorrect:

- Nucleotide excision repair: Repairs bulky DNA lesions such as thymine dimers caused by UV radiation (defective in xeroderma pigmentosum).
- Base excision repair: Corrects small base modifications such as deamination or oxidation.
- Double-strand break repair: Repairs DNA breaks through homologous recombination or non-homologous end joining.

QUESTION

| When a codon is changed to stop codon, then it is:

A Stop codon mutation

B **Non-sense mutation**

CORRECT

C Mis sense Mutation

D Silent Mutation

RIGHT ANSWER

OK **B. Non-sense mutation**

WHY THIS IS RIGHT

A nonsense mutation is a type of point mutation in which a codon for an amino acid is converted into a stop codon (UAA, UAG, or UGA). This results in premature termination of protein synthesis, producing a truncated and usually nonfunctional protein.

Why other options are incorrect:

- Stop codon mutation: Not a standard classification term used in genetics.
- Missense mutation: One amino acid is replaced by another (no stop codon formed).
- Silent mutation: Codon change does not alter the amino acid due to redundancy of the genetic code.

QUESTION

| True about telomeres is:

- A** Increased telomere length with ageing
- B** Gradually decreasing length of telomeres with ageing CORRECT
- C** Telomeres have coding sequences
- D** Telomerase is found in somatic cells

RIGHT ANSWER

- OK** **B. Gradually decreasing length of telomeres with ageing**

WHY THIS IS RIGHT

Telomeres are repetitive non-coding DNA sequences (TTAGGG repeats) present at the ends of chromosomes that protect them from degradation and fusion.

With each cell division, due to the end-replication problem, telomeres gradually shorten, leading to:

- Cellular aging (senescence)
- Limited replicative capacity of somatic cells

Telomerase is an enzyme that adds telomeric repeats to chromosome ends, thereby maintaining telomere length. However:

- It is active mainly in germ cells, stem cells, and cancer cells
- It is absent or very low in most somatic cells

Why other options are incorrect:

- Increased telomere length with ageing: Opposite is true.
- Telomeres have coding sequences: They are non-coding repetitive DNA.
- Telomerase is found in somatic cells: Generally absent or minimal in somatic cells.

QUESTION

| DNA packing is done by?

A Histone

CORRECT

B Glycoprotein

C Nucleic acid

D Helicases

RIGHT ANSWER

OK

A. Histone

WHY THIS IS RIGHT

DNA packaging in eukaryotic cells is carried out by histone proteins, which help condense long DNA molecules into a compact, organized structure within the nucleus.

- DNA wraps around histone octamers (2 each of H2A, H2B, H3, H4) forming nucleosomes
- Nucleosomes further coil to form chromatin fibers and ultimately chromosomes

Functions of histones:

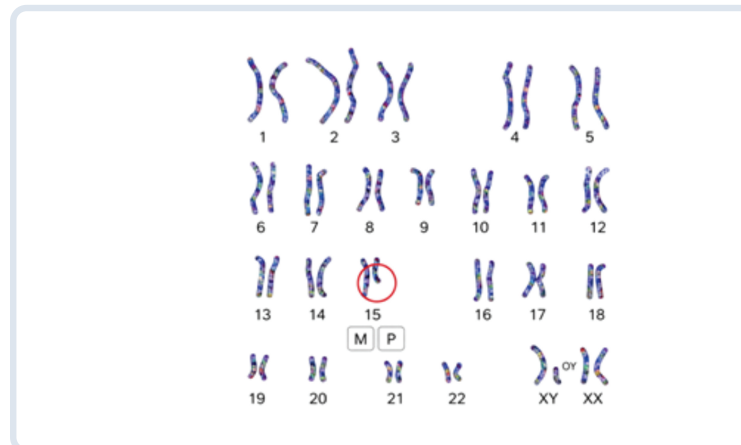
- DNA compaction (fits long DNA into nucleus)
- Regulation of gene expression (via histone modification like acetylation, methylation)

Why other options are incorrect:

- Glycoprotein: Not involved in DNA packaging
- Nucleic acid: DNA itself, not responsible for packing
- Helicases: Enzymes that unwind DNA during replication, not packaging

QUESTION

What is the diagnosis of the marked karyotype?



A Prader-Willi syndrome

CORRECT

B DiGeorge syndrome

C Cri du Chat syndrome

D Angelman syndrome

RIGHT ANSWER

OK A. Prader-Willi syndrome

WHY THIS IS RIGHT

The karyotype shows a defect involving chromosome 15, consistent with Prader-Willi syndrome, caused by deletion of the paternal allele of chromosome 15 (15q11-q13) due to genomic imprinting.

- Paternal gene deletion or maternal uniparental disomy
- Imprinting -> only one parental allele is active
- Loss of paternal gene expression -> disease

Clinical features:

- Hyperphagia -> obesity
- Hypotonia in infancy
- Intellectual disability
- Hypogonadism

Why other options are incorrect:

- DiGeorge syndrome: 22q11 deletion
- Cri du Chat: 5p deletion (cat-like cry)
- Angelman syndrome: Maternal chromosome 15 deletion -> ataxia, seizures, "happy puppet"

QUESTION

A 7-year-old child presents with severe photosensitivity, freckling over sun-exposed areas, and recurrent skin lesions since early childhood. There is a history of progressive neurological decline. On examination, multiple hyperpigmented macules and areas of skin atrophy are noted on the face and arms. The underlying defect involves impaired repair of UV-induced thymine dimers. Which DNA repair mechanism is primarily affected in this condition?

- A** Base excision repair
- B** Double-strand DNA break repair
- C** Mismatch repair

D Nucleotide excision repair

CORRECT

RIGHT ANSWER

OK D. Nucleotide excision repair

WHY THIS IS RIGHT

UV radiation causes DNA damage in the form of pyrimidine (thymine) dimers, which distort the DNA helix. These lesions are repaired by the nucleotide excision repair (NER) pathway.

1. Recognition of DNA distortion (thymine dimers)
2. Excision of the damaged DNA segment
3. DNA polymerase fills the gap
4. DNA ligase seals the strand

Clinical relevance:

- Defect in NER -> Xeroderma pigmentosum (XP)
- Presents with photosensitivity, exaggerated skin rash on sun exposure, and early skin cancers

Why other options are incorrect:

- Base excision repair: Repairs small base changes (oxidation, deamination)
- Double-strand break repair: Repairs DNA breaks (HR, NHEJ)
- Mismatch repair: Corrects replication errors (base mismatches)

QUESTION

A mother brought her 14-year-old mentally retarded child with complaints of joint pain and an irresistible urge to bite his fingers and lips. On investigation, his serum uric acid level was found to be elevated. The child is likely suffering from which disorder?

A Xanthine oxidase deficiency

B Thymidylate synthetase

C Lesch-Nyhan syndrome

CORRECT

D CPSII defect

RIGHT ANSWER

OK C. Lesch-Nyhan syndrome

WHY THIS IS RIGHT

The clinical features-intellectual disability, self-mutilation (biting lips/fingers), joint pain, and hyperuricemia-are characteristic of Lesch-Nyhan syndrome.

- Deficiency of HGPRT (Hypoxanthine-guanine phosphoribosyltransferase)
- A key enzyme in the purine salvage pathway
- Impaired salvage of hypoxanthine and guanine -> increased degradation to uric acid
- ↑ PRPP and ↓ IMP/GMP -> ↑ de novo purine synthesis -> further uric acid production

Clinical features explained:

- Hyperuricemia -> gout, joint pain
- Neurological dysfunction -> intellectual disability
- Self-mutilation behavior (classic hallmark)

Why other options are incorrect:

- Xanthine oxidase deficiency: ↓ uric acid (xanthinuria), not hyperuricemia
- Thymidylate synthetase defect: Affects DNA synthesis
- CPS II defect: Affects pyrimidine synthesis

QUESTION

A frameshift mutation occurs due to the insertion of a new nucleotide at the 4th position of an mRNA sequence containing 900 nucleotides. What is the most likely outcome of this mutation?

A No change in the final protein

B Partial protein production

C Complete change in protein production

CORRECT

D No change due to RNA editing

RIGHT ANSWER

OK **C. Complete change in protein production**

WHY THIS IS RIGHT

A frameshift mutation occurs when nucleotides are inserted or deleted in numbers not divisible by 3, altering the reading frame of the mRNA.

In this case, insertion of a nucleotide at the 4th position (early in the sequence) causes:

- Shift in the reading frame from that point onward
- All subsequent codons are altered
- Often leads to a premature stop codon
- Produces a completely abnormal (nonfunctional) protein

Why other options are incorrect:

- No change in protein: Seen in silent mutations, not frameshift
- Partial protein production: More typical of nonsense mutation (premature stop without frame shift)
- No change due to RNA editing: RNA editing does not correct frameshift mutations

QUESTION

A child presents with multiple freckles, severe photosensitivity, and early-onset skin cancers. What is the primary underlying molecular defect responsible for this condition?

A Nucleotide excision repair defect

CORRECT

B Mismatch repair defect

C Base excision repair defect

D Homologous recombination defect

RIGHT ANSWER

OK

A. Nucleotide excision repair defect

WHY THIS IS RIGHT

The clinical features-multiple freckles, severe photosensitivity, and early-onset skin cancers-are characteristic of Xeroderma Pigmentosum (XP).

Underlying molecular defect:

- Defective nucleotide excision repair (NER) pathway
- This pathway normally repairs UV-induced DNA damage, especially pyrimidine (thymine) dimers

Pathophysiology:

- UV radiation -> formation of thymine dimers
- NER defect -> inability to remove damaged DNA
- Accumulation of mutations -> high risk of skin cancers at an early age

Why other options are incorrect:

- Mismatch repair defect: Seen in Lynch syndrome (HNPCC)
- Base excision repair defect: Repairs small base damage (oxidation, deamination)
- Homologous recombination defect: Seen in BRCA mutations (breast/ovarian cancer)

QUESTION

Which PCR technique is considered the most appropriate method for syndromic diagnosis of pathogens causing meningitis?

- A

Nested PCR
- B

Multiplex PCR

CORRECT
- C

RT-PCR
- D

Uniplex PCR

RIGHT ANSWER

OK

B. Multiplex PCR

WHY THIS IS RIGHT

No explanation was provided in the source quiz.

PCR Type	Location	Function
Multiplex PCR		Uses multiple primer sets in one reaction to detect several pathogens simultaneously.
Nested PCR		Two-step PCR using inner + outer primers to give high sensitivity/specificity for a single low-copy organism.
RT-PCR (qPCR)		Real-time fluorescent PCR giving quantitative detection of one target per reaction (unless multiplexed).
Uniplex PCR		Standard PCR that detects only one organism per reaction, needing separate tubes for each target.

QUESTION

| Which technique is used to identify auto antibodies?

A Direct immunofluorescence

B ELISA

C Indirect immunofluorescence

CORRECT

D FISH

RIGHT ANSWER

OK C. Indirect immunofluorescence

WHY THIS IS RIGHT

Indirect immunofluorescence (IIF) is commonly used to detect circulating autoantibodies in patient serum. In this technique, the patient's serum is incubated with a substrate containing the target antigen (e.g., cells or tissue). If autoantibodies are present, they bind to the antigen. A fluorescent-labeled secondary antibody (anti-human IgG) is then added to detect these bound antibodies, producing fluorescence under a microscope.

Why other options are incorrect:

- Direct immunofluorescence: Detects antigen-antibody complexes deposited in tissues, not circulating autoantibodies in serum.
- ELISA: Can detect antibodies, but the classical and widely used screening method for many autoantibodies (e.g., ANA patterns) is indirect immunofluorescence.
- FISH (Fluorescence in situ hybridization): Used to detect DNA or chromosomal abnormalities, not antibodies.

QUESTION

In which of the genetic material, Number of Adenine will not be equal to that of Thymine:

- A** E. coli
- B** Mitochondrial DNA of human
- C** HIV
- D** Herpes virus

CORRECT

RIGHT ANSWER

OK C. HIV

WHY THIS IS RIGHT

According to Chargaff's rule, in double-stranded DNA the amount of adenine (A) equals thymine (T) because they pair together (A-T). This rule applies to organisms with double-stranded DNA genomes, such as E. coli, human mitochondrial DNA, and herpes virus.

HIV, however, contains a single-stranded RNA genome, not double-stranded DNA. Since base pairing is not fixed in single-stranded nucleic acids, adenine does not necessarily equal thymine (or uracil). Therefore, Chargaff's rule does not apply, making HIV the correct answer.

QUESTION

Which Biochemical method is exploited for monoclonal antibody production by hybridoma technique?

- A** Glycolytic pathway
- B** Pentose Phosphate pathway
- C** Kreb's cycle
- D** Purine salvage pathway

CORRECT

RIGHT ANSWER

- OK** **D. Purine salvage pathway**

WHY THIS IS RIGHT

The hybridoma technique for producing monoclonal antibodies uses HAT medium (Hypoxanthine-Aminopterin-Thymidine) for selection. In this medium, cells must use the purine salvage pathway to synthesize nucleotides because aminopterin blocks the de novo purine synthesis pathway.

Only hybridoma cells (fusion of antibody-producing B cells with myeloma cells) survive, as they possess the necessary enzymes (e.g., HGPRT) to utilize the purine salvage pathway, allowing selective growth and production of monoclonal antibodies.

Why not other options:

- Glycolytic pathway: Involved in glucose breakdown for ATP, not nucleotide synthesis or HAT selection.
- Pentose phosphate pathway: Produces NADPH and ribose-5-phosphate, but does not bypass the aminopterin block in HAT medium.
- Krebs cycle: Part of energy metabolism in mitochondria, unrelated to nucleotide salvage used in hybridoma selection.

QUESTION

FISH is used to detect all of the following except?

A Translocation

B Amplification

C Deletion

D Point mutation

CORRECT

RIGHT ANSWER

OK **D. Point mutation**

WHY THIS IS RIGHT

Fluorescence in situ hybridization (FISH) is a cytogenetic technique that uses fluorescent DNA probes to detect large chromosomal abnormalities such as translocations, deletions, duplications, and gene amplifications. However, FISH cannot detect point mutations because these involve single nucleotide changes, which are too small to be identified by probe-based chromosomal hybridization. Detection of point mutations requires molecular techniques such as PCR or DNA sequencing.

QUESTION

| Which of the following is not associated with post-transcriptional modification?

- A** Methylation
- B** Endonuclease cleavage
- C** Capping
- D** Glycosylation

CORRECT

RIGHT ANSWER

- OK** **D. Glycosylation**

WHY THIS IS RIGHT

Post-transcriptional modifications occur after RNA is synthesized from DNA and are essential for converting pre-mRNA into mature mRNA. The major modifications include:

- 5' Capping: Addition of a 7-methylguanosine cap at the 5' end, which protects mRNA from degradation and helps in ribosome binding.
- Endonuclease cleavage and splicing: Removal of introns and joining of exons to form mature mRNA.
- Methylation: Certain bases in RNA may be methylated during RNA processing.

Glycosylation is not a post-transcriptional modification. It is a post-translational modification of proteins that occurs in the endoplasmic reticulum and Golgi apparatus, where carbohydrate groups are added to proteins.

QUESTION

Which type of mutation results in the reversal to the wild type of phenotype when the mutant gene is suppressed?

A Frameshift mutation of coding gene

B Addition of another normal gene

C Deletion of the mutant gene

D Mutation of tRNA

CORRECT

RIGHT ANSWER

OK D. Mutation of tRNA

WHY THIS IS RIGHT

A suppressor mutation can restore the wild-type phenotype of a mutant gene without correcting the original mutation. One common mechanism involves a mutation in tRNA that allows it to recognize altered codons (such as stop codons or frameshift codons) and insert an amino acid during translation.

These mutant tRNAs (suppressor tRNAs) compensate for the defective coding sequence, permitting translation to continue and partially or completely restoring protein function.

Thus, mutation of tRNA can suppress the effect of the original mutation and revert the phenotype toward wild type, even though the original DNA mutation remains present.

QUESTION

Which of the following is the best investigation for aneuploidy?

- A. In Situ Hybridization
- B. Conventional cytogenetics
- C. PCR
- D. Sanger sequencing

A A only

B A & B

CORRECT

C C & D

D A, B, C and D

RIGHT ANSWER

OK B. A & B

WHY THIS IS RIGHT

Aneuploidy refers to an abnormal number of chromosomes (e.g., trisomy or monosomy). The most appropriate investigations are karyotyping (conventional cytogenetics) and fluorescence in situ hybridization (FISH) because they directly evaluate chromosome number and structure.

- Conventional cytogenetics (karyotyping) visualizes the entire chromosome set and is the gold standard for detecting numerical abnormalities such as trisomy 21, trisomy 18, and monosomy X.
- In situ hybridization (FISH) uses fluorescent probes targeting specific chromosomes and provides rapid detection of aneuploidy, commonly used in prenatal and interphase cells.

PCR and Sanger sequencing are primarily used to detect gene-level mutations or small sequence variations, not whole-chromosome number abnormalities. Therefore, they are not ideal for detecting aneuploidy.

QUESTION

| All are used in recombinant DNA technology (genetic engineering) except?

A Phosphatases

B CRISPR Cas9

C **Isomerases**

CORRECT

D Terminal transferase

RIGHT ANSWER

OK **C. Isomerases**

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$19” here and does not include a written explanation for this item.

QUESTION

A scientist is culturing a virus using the RT-PCR technique and performs 26 amplification cycles. After how many cycles would the DNA quantity be approximately half of what is present at the 26th cycle?

A 27

B 25

CORRECT

C 13

D 10

RIGHT ANSWER

OK B. 25

WHY THIS IS RIGHT

In PCR amplification, the amount of DNA doubles with each cycle (exponential amplification). the DNA quantity at 25 cycles is approximately half of that at 26 cycles.

$$\text{DNA amount} = 2^n$$

• At 26 cycles $\rightarrow 2^{26}$ copies

• At 25 cycles $\rightarrow 2^{25}$ copies

Since

$$2^{25} = \frac{1}{2} \times 2^{26}$$

QUESTION

Match the following:

Column 1	Column 2
1. Southern blotting	a. Proteins
2. Northern blotting	b. Enzymes
3. Western blotting	c. RNA
4. Eastern blotting	d. DNA

A 1-a, 2-d, 3-c, 4-b

B 1-d, 2-c, 3-a, 4-b

CORRECT

C 1-a, 2-b, 3-c, 4-d

D 1-b, 2-c, 3-d, 4-a

RIGHT ANSWER

OK B. 1-d, 2-c, 3-a, 4-b

WHY THIS IS RIGHT

No explanation was provided in the source quiz.

Technique	Detects	Key Use
Southern blot	DNA	Gene mapping, detecting mutations, RFLP analysis, prenatal diagnosis
Northern blot	RNA	Measures gene expression (mRNA levels), differentiates active vs. silent genes
Western blot	Proteins	Confirms HIV , Lyme disease, detects specific antibodies/proteins
Eastern blot	Enzymes/post-translational modifications (e.g., glycosylation)	Used to study protein modifications important in signaling pathways

QUESTION

Which of the following is not an example of aneuploidy?

A Klinefelter syndrome

B Patau syndrome

C Down syndrome

D Bloom syndrome

CORRECT

RIGHT ANSWER

OK **D. Bloom syndrome**

WHY THIS IS RIGHT

Aneuploidy refers to an abnormal number of chromosomes due to nondisjunction during meiosis, resulting in gain or loss of individual chromosomes.

Examples of Aneuploidy

- Down syndrome: Trisomy 21
- Patau syndrome: Trisomy 13
- Klinefelter syndrome: 47, XXY (sex chromosome aneuploidy)

Bloom syndrome is not an aneuploidy; it is a chromosomal instability disorder caused by mutation in the BLM gene (DNA helicase) leading to defective DNA repair and increased sister chromatid exchanges.

QUESTION

A 58-year-old patient with a known malignancy undergoes molecular analysis of tumor cells. RNA sequencing reveals transcripts that are longer than normal, with inclusion of additional exon sequences not seen in healthy tissue. Which of the following mechanisms best explains this finding?

- A** Polyadenylation
- B** Alternative RNA splicing
- C** Gene amplification
- D** Upstream promoter modification

CORRECT

RIGHT ANSWER

- OK** **B. Alternative RNA splicing**

WHY THIS IS RIGHT

Alternative RNA splicing allows a single gene to produce multiple mRNA transcripts with different exon combinations, leading to variation in transcript length and protein isoforms.

Role in Cancer:

- Cancer cells often exhibit abnormal alternative splicing.
- This can generate longer or altered mRNA transcripts.
- These variants may promote tumor growth, survival, and metastasis.

Why not other options:

- A) Polyadenylation: Adds a poly-A tail at the 3' end but does not significantly change transcript length of coding regions.
- C) Gene amplification: Increases the number of gene copies, not the length of each RNA transcript.
- D) Upstream promoter modification: Affects transcription initiation and expression level, not mRNA length.

QUESTION

A researcher treats cultured human cells with a histone acetyltransferase (HAT) inhibitor and observes rapid epigenetic changes. Within minutes, there is reduced transcriptional activity and chromatin appears more compact. Which of the following molecular changes is most likely to be seen immediately?

A Increased H3K9me2/3

CORRECT

B Increased H3K9 acetylation

C Chromatin relaxation

D DNA methylation

RIGHT ANSWER

OK

A. Increased H3K9me2/3

WHY THIS IS RIGHT

- HAT inhibition → ↓ histone acetylation → chromatin condensation
- Associated with repressive marks (↑ H3K9 methylation)
- Acetylation = active chromatin; methylation (H3K9me2/3) = silenced chromatin

Histone acetylation relaxes chromatin and promotes gene transcription, whereas histone deacetylation leads to chromatin condensation and gene repression.

Effect of Histone Acetylase (HAT) Inhibition:

- ↓ Histone acetylation
- Chromatin becomes more condensed
- Leads to repressive histone marks, such as H3K9 methylation (H3K9me2/3)

QUESTION

Which of the following correctly depicts the sequence of steps in the PCR?

A Denaturation -> Annealing -> Extension

CORRECT

B Denaturation -> Annealing -> Extension -> Hybridization

C Annealing -> Extension -> Denaturation

D Extension -> Denaturation -> Annealing

RIGHT ANSWER

OK A. Denaturation -> Annealing -> Extension

WHY THIS IS RIGHT

Polymerase Chain Reaction (PCR) is a laboratory technique used to amplify a specific segment of DNA, producing millions of copies from a small DNA sample.

Sequence of steps in PCR

1. Denaturation ($\approx 94-98^{\circ}\text{C}$)
 - Double-stranded DNA separates into two single strands due to breaking of hydrogen bonds.
2. Annealing ($\approx 50-65^{\circ}\text{C}$)
 - Primers bind to complementary sequences on the single-stranded DNA template.
3. Extension / Elongation ($\approx 72^{\circ}\text{C}$)
 - DNA polymerase synthesizes new DNA strands by adding nucleotides complementary to the template.

These steps are repeated for multiple cycles, resulting in exponential amplification of the target DNA.

QUESTION

Researchers are working to identify new targets for drug development to treat idiopathic pulmonary fibrosis. After obtaining lung tissue samples from affected patients, they conduct a series of experiments to measure the expression and activity of various profibrotic proteins. The results for one such protein, galectin-3, are as follows:

mRNA concentration: Normal

Protein concentration: Increased

Protein activity: Intact

Which of the following processes best explains these findings?

A Mutation in the Galectin-3 promoter region

B Decreased methylation of the Galectin-3 gene

C Shortening of the Galectin-3 mRNA poly-(A) tail

D Decreased ubiquitination of the Galectin-3 protein

CORRECT

RIGHT ANSWER

OK D. Decreased ubiquitination of the Galectin-3 protein

WHY THIS IS RIGHT

The patient's lung tissue shows normal galectin-3 mRNA but increased protein levels with intact activity, indicating that transcription is normal but protein degradation is reduced. Ubiquitination tags proteins for proteasomal degradation, so decreased ubiquitination stabilizes galectin-3, leading to higher protein levels without changing mRNA or activity.

Why the others are incorrect:

A. Mutation in the Galectin-3 promoter region: Would increase mRNA expression, which is normal here.

B. Decreased methylation of the Galectin-3 gene: Typically increases transcription, raising mRNA levels, not observed.

C. Shortening of the Galectin-3 mRNA poly-(A) tail: Usually reduces mRNA stability, leading to lower protein, opposite of what is seen.

QUESTION

An investigator is studying the genotypes of fire-flies using full exome sequencing. They are found to have a point mutation in the gene encoding wing bud formation during embryogenesis. The point mutation in the gene causes the mRNA transcript to have a "UUG" segment instead of an "AUG" segment. Which of the following processes is most likely affected by this mutation?

- A** Cleavage of 5' intron
- B** Catalyzation of peptide bond formation
- C** Binding of met-tRNA to 60S complex
- D** Dissociation of mRNA from ribosome complex

CORRECT

RIGHT ANSWER

- OK** **C. Binding of met-tRNA to 60S complex**

WHY THIS IS RIGHT

The point mutation changes the start codon from AUG -> UUG. AUG is the canonical start codon that recruits initiator methionine tRNA (Met-tRNA_i) to the small ribosomal subunit (40S in eukaryotes) and subsequently forms the 60S/40S initiation complex. A non-AUG codon prevents proper Met-tRNA binding, inhibiting translation initiation of the mRNA.

Why the others are incorrect:

- A. Cleavage of 5' intron: This occurs during mRNA splicing, which is upstream of translation and unaffected by start codon mutation.
- B. Catalyzation of peptide bond formation: This occurs at the ribosome during elongation; the start codon mutation prevents initiation, so elongation never occurs.
- D. Dissociation of mRNA from ribosome complex: Happens during translation termination, which is downstream of initiation and unaffected by the start codon mutation.

QUESTION

A molecular biologist is working on isolating a specific gene within a large DNA fragment ranging from 50 to 100 kilobases (KB) in length. Which technique is most suitable for this purpose?

A Notch

B RFLP

C SSLP

D Chromosome walking

CORRECT

RIGHT ANSWER

OK

D. Chromosome walking

WHY THIS IS RIGHT

Chromosome walking is a technique used to isolate and sequence a gene within large DNA fragments (tens to hundreds of kilobases). It involves progressive cloning and sequencing of overlapping DNA fragments starting from a known marker or sequence, allowing researchers to “walk” along the chromosome to reach the gene of interest.

Why the others are incorrect:

A. Notch: Not a DNA isolation method; it refers to a signaling pathway or gene.

B. RFLP (Restriction Fragment Length Polymorphism): Detects DNA polymorphisms but is not a method for isolating large genes.

C. SSLP (Simple Sequence Length Polymorphism): Used for genetic mapping; it does not help clone long DNA sequences.

QUESTION

| Which of the following is most directly responsible for triplex DNA formation?

- A** Hoogsteen pairing
- B** Palindromic sequences
- C** Large no. of guanosine repeats
- D** Polypyrimidine tracts

CORRECT

RIGHT ANSWER

- OK** **A. Hoogsteen pairing**

WHY THIS IS RIGHT

Triplex DNA forms when a third strand of DNA binds to the major groove of a double-stranded DNA through Hoogsteen hydrogen bonds. This usually occurs in polypurine-polypyrimidine regions, allowing the third strand to bind parallel or antiparallel to the purine-rich strand. Hoogsteen pairing is the specific hydrogen-bonding mechanism that stabilizes triplex formation.

Why the others are incorrect:

- B. Palindromic sequences: Facilitate hairpin or cruciform structures, not triplex DNA.
- C. Large number of guanosine repeats: Can form G-quadruplexes, not classical triplex DNA.
- D. Polypyrimidine tracts: Often present in triplex-prone regions but do not directly cause triplex formation; Hoogsteen pairing is required for stabilization.

QUESTION

Which of the following sequences represents the correct order of enzyme activity in DNA replication?

A DNA polymerase → protein unwinding enzyme → DNA ligase → DNA Isomerase → Polymerase I

B RNA polymerase → DNA polymerase III → DNA polymerase I → DNA ligase

CORRECT

C RNA polymerase → DNA polymerase III → DNA ligase → exonuclease → DNA polymerase I

D Protein unwinding enzyme → polymerase I → DNA ligase → DNA isomerase → DNA polymerase

RIGHT ANSWER

OK B. RNA polymerase → DNA polymerase III → DNA polymerase I → DNA ligase

WHY THIS IS RIGHT

During DNA replication:

1. RNA polymerase (primase) synthesizes a short RNA primer to initiate replication.
2. DNA polymerase III extends the new DNA strand from the RNA primer.
3. DNA polymerase I removes the RNA primer and fills in the gap with DNA.
4. DNA ligase seals the remaining nicks between Okazaki fragments on the lagging strand.

Why the others are incorrect:

- Options starting with DNA polymerase before unwinding ignore the need for helicase/primase first.
- Ordering exonuclease or DNA isomerase incorrectly in the sequence does not match the actual replication process.

QUESTION

If there are 4 consecutive nucleotides code for an amino acid. Then how many amino acid can be coded by nucleic acid?

A 256

CORRECT

B 64

C 16

D 4

RIGHT ANSWER

OK

A. 256

WHY THIS IS RIGHT

Each nucleotide in DNA or RNA can be one of 4 bases (A, T/U, G, C). If 4 consecutive nucleotides code for one amino acid, the total number of possible combinations is: $4^4 = 256$ different codons can theoretically code for amino acids.

Why the others are incorrect:

B. 64: This is the number of codons if 3 nucleotides per amino acid (standard genetic code: $4^3 = 64$).

C. 16: Would be 2 nucleotides per amino acid ($4^2 = 16$).

D. 4: Only 1 nucleotide per amino acid ($4^1 = 4$).

QUESTION

A molecular biologist is preparing to amplify a specific DNA segment using an enzyme catalysed thermocycler-based reaction. Which of the following components if not included in reaction mixture will not affect the amplification process?

A

DNA ligase

CORRECT

B

Taq polymerase

C

DNA primers & Magnesium ions (Mg^{2+})

D

dNTPs (deoxynucleotide triphosphates)

RIGHT ANSWER

OK

A. DNA ligase

WHY THIS IS RIGHT

In PCR (polymerase chain reaction), amplification requires Taq polymerase, DNA primers, Mg^{2+} ions, and dNTPs. DNA ligase is not required, because PCR synthesizes new DNA strands without joining separate DNA fragments; ligase only seals nicks in DNA, which is unnecessary for standard PCR amplification.

Why the others are incorrect:

B. Taq polymerase: Catalyzes DNA synthesis; without it, no amplification occurs.

C. DNA primers & Mg^{2+} ions: Primers provide initiation sites for polymerase; Mg^{2+} is a cofactor essential for enzyme activity.

D. dNTPs: Serve as building blocks for new DNA strands; omission halts synthesis.

QUESTION

Which of the following acts as an RNA-directed nuclease?

A CRISPR-Cas9

CORRECT

B S1 nuclease

C Primase

D Telomerase

RIGHT ANSWER

OK **A. CRISPR-Cas9**

WHY THIS IS RIGHT

The CRISPR-Cas9 system uses a guide RNA to direct the Cas9 nuclease to a specific DNA sequence, allowing targeted cleavage. Although Cas9 itself is a DNA endonuclease, its activity is RNA-directed, meaning the RNA sequence determines where the nuclease cuts. This mechanism underlies gene editing and genome engineering applications.

Why the others are incorrect:

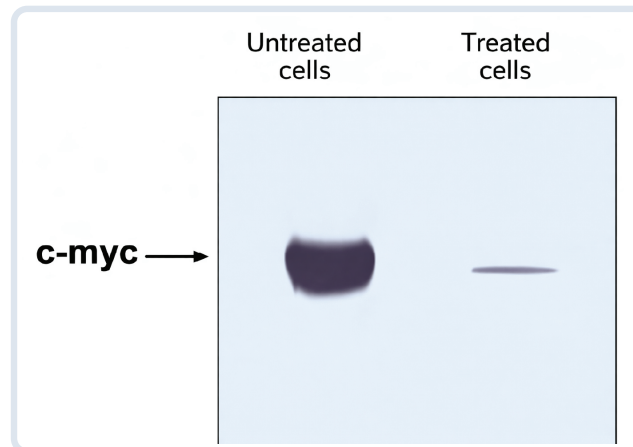
B. S1 nuclease: Cleaves single-stranded DNA or RNA but is not guided by RNA.

C. Primase: Synthesizes RNA primers for DNA replication; it does not cleave nucleic acids.

D. Telomerase: Adds repetitive DNA sequences to chromosome ends using its RNA template, but it functions as a reverse transcriptase, not an RNA-directed nuclease.

QUESTION

A pharmaceutical corporation investigating new therapeutic agents for treatment of Burkitt lymphoma synthesizes a double-stranded RNA molecule that is 21 base pairs in length. The RNA molecule has a base pair sequence that is complementary to a region of mRNA encoding c-Myc. Introduction of the RNA molecule into tumor cells results in a significant reduction in cell growth. Western blot analysis of equivalent numbers of treated and untreated cells is shown below. Which of the following processes was most likely directly interrupted in the cells exposed to the RNA molecule?



A DNA replication

B DNA transcription

C mRNA translation

CORRECT

D Proteasome activity

RIGHT ANSWER

OK C. mRNA translation

WHY THIS IS RIGHT

The experiment describes small interfering RNA (siRNA) complementary to c-Myc mRNA, which binds the target mRNA and induces its degradation via the RNA-induced silencing complex (RISC). This prevents translation of c-Myc protein, reducing protein levels and inhibiting cell proliferation in Burkitt lymphoma cells.

Why the others are incorrect:

A. DNA replication: The siRNA does not affect DNA synthesis; it targets mRNA.

B. DNA transcription: Transcription proceeds normally; the RNA interference acts post-transcriptionally.

D. Proteasome activity: Protein degradation is not directly inhibited; reduced c-Myc results from decreased translation, not enhanced degradation.

QUESTION

A 2-year-old boy is brought to the emergency department with fever, vomiting, and sleepiness. Since the newborn period, the parents say that the patient has had multiple similar episodes. Lab results revealed increased blood ammonia levels during these episodes and markedly increased orotic acid excretion in the urine. Physical examination shows a tachypneic boy who is unresponsive to all stimuli. Which of the following enzymes is most likely to be deficient in this patient?

- A** Carbamoyl phosphate synthetase I
- B** Hypoxanthine-guanine phosphoribosyltransferase
- C** Ornithine transcarbamylase
- D** Uridine monophosphate synthetase

CORRECT

RIGHT ANSWER

- OK** **C. Ornithine transcarbamylase**

WHY THIS IS RIGHT

This patient presents with hyperammonemia and markedly increased urinary orotic acid, which are hallmark features of ornithine transcarbamylase deficiency, an X-linked urea cycle disorder. OTC deficiency leads to accumulation of carbamoyl phosphate, which is shunted into pyrimidine synthesis, producing excess orotic acid. Clinical presentation includes recurrent vomiting, lethargy, and tachypnea in early childhood.

Why the others are incorrect:

- A. Carbamoyl phosphate synthetase I: Deficiency also causes hyperammonemia but orotic acid is low, not elevated.
- B. Hypoxanthine-guanine phosphoribosyltransferase (HGPRT): Deficiency causes Lesch-Nyhan syndrome (hyperuricemia, self-mutilation), not hyperammonemia.
- D. Uridine monophosphate synthetase: Deficiency leads to orotic aciduria without hyperammonemia; differs from OTC deficiency.

QUESTION

E. coli colonies grown on a lactose-containing medium up-regulate the production of the enzymes β -galactosidase and galactoside permease. Which of the following best explains the synchronous production of both enzymes in response to lactose?

A There are two activator binding sites for one activator protein.

B There are two repressors for one inducer.

C There are two promoters near each other.

D There is one mRNA coding for both enzymes.

CORRECT

RIGHT ANSWER

OK **D. There is one mRNA coding for both enzymes.**

WHY THIS IS RIGHT

The lac operon is a classic example of polycistronic mRNA, where a single mRNA transcript encodes multiple enzymes, including β -galactosidase and galactoside permease. This allows synchronous expression of all enzymes needed to metabolize lactose in response to the inducer.

Why the others are incorrect:

A. Two activator binding sites: Not required; regulation is mainly via the lac repressor and CAP.

B. Two repressors for one inducer: Only the lac repressor regulates operon induction.

C. Two promoters: There is a single promoter driving the polycistronic transcript; multiple promoters are unnecessary for synchronized expression.

QUESTION

In an animal experiment, mice proerythroblasts are cultured in 2 different growth media; the first medium is folate deficient, whereas the second (control) is supplemented with folic acid. Both media contain high concentrations of erythropoietin. Over 48 hours, cells in the control media proliferate and differentiate into reticulocytes, whereas in the folate-deficient media, cell proliferation is minimal, with most cells undergoing apoptosis. In another experiment, a substance is added to the folate-deficient media, which prevents apoptosis and permits proliferation of the proerythroblasts. Which of the following is the most likely substance added to the growth medium?

A Cobalamin

B Cytosine

C Homocysteine

D Thymidine

CORRECT

RIGHT ANSWER

OK D. Thymidine

WHY THIS IS RIGHT

Folate deficiency impairs thymidylate (dTMP) synthesis, leading to inability to synthesize DNA, cell cycle arrest, and apoptosis in rapidly dividing proerythroblasts. Supplementing thymidine, a pyrimidine nucleoside, bypasses the block in DNA synthesis, allowing DNA replication and proliferation even without folate.

Why the others are incorrect:

- A. Cobalamin (Vitamin B12): Required for folate recycling, but supplementation alone cannot bypass dTMP deficiency.
- B. Cytosine: A nucleobase that cannot be directly incorporated into DNA; does not rescue proliferation.
- C. Homocysteine: Accumulates in folate deficiency; adding it does not restore DNA synthesis.

QUESTION

A 34-year-old woman comes to the OPD after her sister was diagnosed with breast cancer. Her sister was found to have a multiple base pair insertion affecting exon 11 of the BRCA 1 gene that leads to a frameshift mutation. A screening test to evaluate for a similar insertion mutation in the patient's BRCA 1 gene is performed. The test uses polymerase chain reaction (PCR) to amplify the target exon and gel electrophoresis to assess the size of the exon compared to the wild-type allele. Which of the following must be known to perform the amplification part of this analysis?

A Restriction enzyme susceptibility sites within the target exon

B The amino acid sequence of the abnormal BRCA 1 protein

C The complete nucleotide sequence of the target exon

D The nucleotide sequence of the regions flanking the target exon.

CORRECT

RIGHT ANSWER

OK D. The nucleotide sequence of the regions flanking the target exon.

WHY THIS IS RIGHT

PCR amplification requires primers that bind to sequences flanking the target exon. Knowing these flanking sequences allows the primers to anneal specifically, enabling amplification of the exon containing the potential insertion mutation. After amplification, gel electrophoresis can detect size changes indicative of insertions or deletions.

Why the others are incorrect:

A. Restriction enzyme sites: Only needed for restriction fragment analysis, not standard PCR.

B. Amino acid sequence: Protein sequence cannot be used to design DNA primers; DNA sequence is needed.

C. Complete nucleotide sequence of exon: Knowing the exon alone is insufficient; primers must bind to flanking regions for PCR amplification.

QUESTION

15-year-old boy is found to have unexplained erythrocytosis on routine laboratory analysis. Evaluation of his immediate family shows that his father and sister also have elevated red cell levels. Genetic sequencing of the 13-globin gene is performed in the affected family members. The results showed a single base substitution at amino acid position 82 that replaces the normal lysine residue with methionine. Further analysis shows that this amino acid replacement impairs the ionic interaction between the 13-subunit and 2,3-bisphosphoglycerate. As a result of this mutation, the patient's haemoglobin will be most like which of the following haemoglobin types?

A Haemoglobin A1c

B Haemoglobin C

C Haemoglobin F

CORRECT

D Haemoglobin S

RIGHT ANSWER

OK C. Haemoglobin F

WHY THIS IS RIGHT

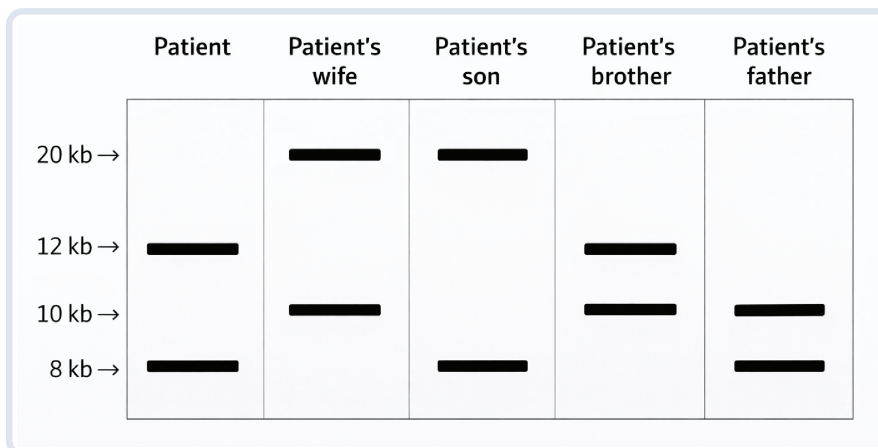
The patient shows familial erythrocytosis due to a mutation in the β -globin gene that increases hemoglobin's oxygen affinity by impairing 2,3-BPG binding. High-affinity hemoglobins (like HbF) bind oxygen more tightly, reducing oxygen delivery to tissues, which stimulates erythropoiesis, explaining the erythrocytosis. The amino acid substitution increases oxygen affinity, mimicking the functional effect of fetal hemoglobin (HbF).

Why the others are incorrect:

- A. Hemoglobin A1c: Formed by glycation of normal HbA in hyperglycemia; unrelated to genetic substitutions.
- B. Hemoglobin C: Lys82 mutation in β -globin causes mild hemolysis and crystal formation, but does not cause high-affinity hemoglobin or erythrocytosis.
- D. Hemoglobin S: Glu6->Val mutation causes sickling, hemolysis, and vaso-occlusive crises, not erythrocytosis.

QUESTION

A 34-year-old man is found to have an LDL level of 310 mg/dl and a normal serum triglyceride level. His father suffered a myocardial infarction at age 39, and his paternal grandfather died of a heart attack at age 40. The patient's wife has a normal lipid profile. DNA samples are obtained from several family members for genetic analysis. Southern blotting of restriction fragments from a region containing the LDL receptor gene shows the following pattern. Identify the correct statement:



A The disease is transmitted in an X-linked recessive fashion.

B The mutation is probably located in the 10 kb band.

C The patient's brother most likely inherited the mutation.

D The patient's son most likely inherited the mutation.

CORRECT

RIGHT ANSWER

OK

D. The patient's son most likely inherited the mutation.

WHY THIS IS RIGHT

This patient has autosomal dominant familial hypercholesterolemia (FH). In AD inheritance, each child has a 50% chance of inheriting the mutated LDL receptor gene. Given the patient is affected, his son is at high risk of inheriting the mutation, making statement D correct.

Why the others are incorrect:

A. X-linked recessive: FH is autosomal dominant, so this inheritance pattern is wrong.

B. Mutation in 10 kb band: The specific band may indicate a polymorphism, but the son's inheritance is more clinically relevant.

C. Patient's brother: Could have inherited it, but we cannot assume without testing; less certain than the son's status.

QUESTION

Which methods among the following can identify changes in the DNA sequence?

1. Restriction fragment length analysis
2. Flow cytometry
3. Pyrosequencing
4. FISH

A 1 and 3

CORRECT

B 1, 2, and 3

C 2, 3, and 4

D 1, 2, and 4

RIGHT ANSWER

OK **A. 1 and 3**

WHY THIS IS RIGHT

Changes in the DNA sequence can be detected by:

- Restriction fragment length analysis, which identifies sequence variations that create or abolish restriction sites.
- Pyrosequencing, a DNA sequencing method that directly determines the nucleotide sequence, allowing mutation detection.

Why the others are incorrect:

2\). Flow cytometry: Analyzes cell size, granularity, and protein expression; it does not detect DNA sequence changes.

4\). FISH (Fluorescence In Situ Hybridization): Detects large chromosomal abnormalities or specific DNA regions but cannot identify small sequence changes.

QUESTION

A microbiologist performs a genetic experiment in which cultures of *Escherichia coli* are treated with a chemical that induces a high frequency of mutations. Individual bacterial colonies are isolated to identify a mutant strain that lacks a specific enzyme involved in DNA replication. This specific enzyme is responsible for removing short fragments of RNA that are base paired to the DNA template. Which of the following enzymes is most likely deficient in this strain of *E. coli*?

A DNA polymerase I

CORRECT

B DNA polymerase III

C Gyrase

D Primase

RIGHT ANSWER

OK A. DNA polymerase I

WHY THIS IS RIGHT

In *E. coli*, DNA polymerase I has both polymerase and 5'→3' exonuclease activity, allowing it to remove RNA primers from Okazaki fragments and replace them with DNA during replication. A deficiency in this enzyme prevents proper removal of RNA primers, leading to incomplete lagging-strand synthesis.

Why the others are incorrect:

- B. DNA polymerase III: Main enzyme for DNA elongation but cannot remove RNA primers.
- C. Gyrase: Relieves supercoiling ahead of the replication fork; unrelated to primer removal.
- D. Primase: Synthesizes RNA primers but does not remove them.

QUESTION

DNA methylation is not related to:

A Mismatch repair

B Chromatin Remodeling

C Capping

CORRECT

D Gene silencing

RIGHT ANSWER

OK C. Capping

WHY THIS IS RIGHT

- DNA methylation involves the addition of methyl groups to cytosine residues, primarily affecting gene expression and chromatin structure.
- It plays a key role in gene silencing, chromatin remodeling, and can influence mismatch repair by affecting recognition of errors during replication.
- The 5'-end capping of mRNA is a post-transcriptional modification unrelated to DNA methylation.

Why the others are incorrect:

- A. Mismatch repair: DNA methylation helps distinguish the parental strand from the newly synthesized strand, aiding repair.
- B. Chromatin remodeling: Methylated DNA recruits proteins that modify chromatin structure, regulating accessibility.
- D. Gene silencing: Methylation at promoter regions represses transcription.

QUESTION

Which of the following methods uses specialized RT-PCR to amplify the ends of mRNA transcripts?

A RACE-PCR

CORRECT

B Multiplex PCR

C Nested PCR

D Droplet digital PCR

RIGHT ANSWER

OK **A. RACE-PCR**

WHY THIS IS RIGHT

RACE-PCR (Rapid Amplification of cDNA Ends) is a specialized RT-PCR technique designed to amplify the 5' or 3' ends of mRNA transcripts. It allows researchers to obtain full-length cDNA sequences from partial mRNA sequences, helping in gene characterization and mapping transcription start/end sites.

Why the others are incorrect:

- B. Multiplex PCR amplifies multiple DNA targets simultaneously in a single reaction, not specifically mRNA ends.
- C. Nested PCR increases specificity by using two successive PCRs, but it does not target transcript ends.
- D. Droplet digital PCR quantifies nucleic acids precisely by partitioning samples into droplets, unrelated to end amplification.

QUESTION

Which of the following site does the restriction enzyme act when RFLP is used in-order to identify the five different species of staphylococci in a surgical ICU?

A ATGGAC - TACGTG

B GATATC - CTATAG

CORRECT

C TAGATA - ATCTAT

D AATATA - TATAAT

RIGHT ANSWER

OK B. GATATC - CTATAG

WHY THIS IS RIGHT

- Restriction Fragment Length Polymorphism (RFLP) analysis relies on restriction enzymes (restriction endonucleases) that recognize specific palindromic DNA sequences and cut at or near these sites.
- GATATC - CTATAG is a palindromic recognition site recognized by the EcoRV restriction enzyme, commonly used in RFLP studies to differentiate bacterial species, including staphylococci.
- Cutting at these specific sites generates DNA fragments of species-specific lengths, which can be resolved by gel electrophoresis to identify different strains.

Why other options are incorrect:

- A. ATGGAC - TACGTG: Not a common restriction site; not palindromic.
- C. TAGATA - ATCTAT: Rare, not typically recognized by standard restriction enzymes.
- D. AATATA - TATAAT: Not a palindromic site for restriction enzymes; resembles a promoter consensus sequence (-10 box in bacteria), not for RFLP.

QUESTION

| Which of the following does not require 5' capping?

A tRNA of Alanine

CORRECT

B U6 SnRNA

C mRNA for Histone

D siRNA

RIGHT ANSWER

OK **A. tRNA of Alanine**

WHY THIS IS RIGHT

5' capping (addition of 7-methylguanosine) occurs on RNA polymerase II transcripts, mainly mRNA and some small nuclear RNAs (snRNAs) like U1, U2, U4, U5, to:

- Protect RNA from degradation
- Facilitate nuclear export
- Promote translation initiation

tRNAs are transcribed by RNA polymerase III and instead undergo processing and modification at their ends, not 5' capping. Similarly, siRNAs are processed from double-stranded RNA and do not require 5' caps. Histone mRNAs are an exception; they have a special 3' stem-loop instead of a poly-A tail, but still receive a 5' cap.

Why other options are incorrect:

- B. U6 snRNA: Does not get a 7-methylguanosine cap, but it is transcribed by RNA polymerase III; still small nuclear RNA function.
- C. mRNA for histone: 5' capping occurs even for histone mRNAs.
- D. siRNA: Not capped, processed from longer dsRNA precursors.

QUESTION

RNA produced from a fragment of DNA has the sequence of UAAGGC. The sequence of the non-template strand in the DNA that gave rise to this sequence is which one of the following?

A CGGAAT

B UAAGGC

C TAAGGC

CORRECT

D ATTCCG

RIGHT ANSWER

OK C. TAAGGC

WHY THIS IS RIGHT

During transcription:

- RNA is synthesized complementary to the DNA template strand.
- The non-template (coding) strand has the same sequence as the RNA, except T replaces U.
- Given RNA: UAAGGC
- Replace U -> T for DNA: TAAGGC

This corresponds to the non-template (coding) strand.

Why other options are incorrect:

- A. CGGAAT: Complement of RNA; this is the template strand, not the coding strand.
- B. UAAGGC: This is RNA, not DNA.
- D. ATTCCG: Incorrect; the first base should be T -> matches U in RNA.

QUESTION

| SARS CoV-2 diagnosis is done by all except:

- A** RT PCR
- B** Real time PCR
- C** Southern blotting
- D** Immunoassays

CORRECT

RIGHT ANSWER

- OK** **C. Southern blotting**

WHY THIS IS RIGHT

SARS-CoV-2 diagnosis relies on detection of viral RNA or viral antigens/antibodies:

- RT-PCR / Real-time PCR: Detect viral RNA with high sensitivity; gold standard for diagnosis.
- Immunoassays: Detect viral antigens (rapid tests) or host antibodies against the virus.
- Southern blotting, however, detects DNA sequences and is not used for RNA viruses like SARS-CoV-2.

Why other options are incorrect:

- A. RT-PCR & B. Real-time PCR: Standard methods for RNA detection.
- D. Immunoassays: Widely used for serological testing or rapid antigen detection.

QUESTION

The base sequence of the strand of DNA used as the template for transcription has the base sequence GATCTAC. What is the base sequence of RNA product?

A CUAGAUG

B GTAGATC

C GAUCUAC

D GUAGAUC

CORRECT

RIGHT ANSWER

OK D. GUAGAUC

WHY THIS IS RIGHT

During transcription, RNA is synthesized complementary to the DNA template strand and in the 5' → 3' direction.

- DNA template strand: GATCTAC (3' → 5')
- RNA transcript: GAUCUAC (5' → 3'), complementary to the DNA template (A → U, T → A, G → C, C → G).

Why other options are incorrect:

- A. CUAGAUG: Reverse complement, not correct orientation.
- B. GTAGATC: Same as DNA template; RNA is complementary, not identical.
- C. GUAGAUC: Wrong base pairing; T → A, not U → G.

QUESTION

The attachment of Eukaryotic mRNA to the ribosome initiation factor is mediated through:

- A Poly A tail
- B tRNA
- C 5'methylGuanyl Cap**
- D Shine-Dalgarno Sequence

CORRECT

RIGHT ANSWER

C. 5'methylGuanyl Cap

WHY THIS IS RIGHT

In eukaryotes, the 5' cap (7-methylguanosine) of mRNA is recognized by eukaryotic initiation factors (eIFs), which mediate the attachment of the mRNA to the ribosome. This cap-dependent mechanism ensures proper ribosome binding and scanning for the start codon (AUG).

Why other options are incorrect:

- A. Poly-A tail: Stabilizes mRNA and aids in nuclear export but does not directly mediate ribosome attachment.
- B. tRNA: Delivers amino acids during elongation, not involved in ribosome binding of mRNA.
- D. Shine-Dalgarno sequence: Found in prokaryotic mRNA, aligns ribosome with start codon; absent in eukaryotes.

QUESTION

Which of the following statements is incorrect about histones?

- A** Histones are synthesized during S phase of the cell cycle
- B** H1 histone does not contribute to formation of nucleosome
- C** Mitochondrial DNA has histone proteins
- D** Histones are rich in arginine and lysine

CORRECT

RIGHT ANSWER

- OK** **C. Mitochondrial DNA has histone proteins**

WHY THIS IS RIGHT

Histones are nuclear proteins, and mitochondrial DNA does not contain histones; it is organized with other proteins like TFAM.

Why other options are correct:

- A. Histones synthesized during S phase: True; synchronized with DNA replication.
- B. H1 histone does not contribute to nucleosome formation: True; H1 is a linker histone stabilizing DNA between nucleosomes, not part of the core octamer.
- D. Histones rich in arginine and lysine: True; these positively charged amino acids facilitate binding to negatively charged DNA.

QUESTION

You are expected to produce a protein of interest. Arrange the following steps in a sequence.

- a. Lysis of bacterial cell
- b. Incorporating gene of interest into bacteria
- c. SDS-PAGE
- d. Hybridization
- e. Protein elution/extraction
- f. Expression of protein

A a-b-c-d-e-f

B b-c-d-a-f-e

C b-d-a-c-e-f

D b-f-a-c-d-e

CORRECT

RIGHT ANSWER

OK D. b-f-a-c-d-e

WHY THIS IS RIGHT

The correct sequence for producing a protein of interest is:

1. b - Incorporating gene of interest into bacteria: The target gene is cloned into an expression vector.
2. f - Expression of protein: Bacteria are induced to express the recombinant protein.
3. a - Lysis of bacterial cell: Cells are broken open to release the protein.
4. c - SDS-PAGE: Used to analyze protein expression and estimate size.
5. d - Hybridization: Can confirm the presence of the correct protein or tag (e.g., Western blotting with a probe).
6. e - Protein elution/extraction: Purification of the protein of interest for downstream applications.

QUESTION

Gene repair by CRISPR-Cas9 can occur by all of the following methods except:

- A** Non- Homologous End Joining
- B** Microhomology-mediated end joining
- C** Mismatch repair
- D** Single-strand annealing

CORRECT

RIGHT ANSWER

- OK** **C. Mismatch repair**

WHY THIS IS RIGHT

CRISPR-Cas9 introduces double-strand breaks (DSBs) in DNA. The cell repairs these DSBs via:

- Non-homologous end joining (NHEJ): Direct ligation of DNA ends; often error-prone.
- Microhomology-mediated end joining (MMEJ): Uses short homologous sequences to repair DSBs.
- Single-strand annealing (SSA): Uses repeated sequences flanking the break to repair.

Mismatch repair (MMR), on the other hand, corrects base mispairings after DNA replication and does not repair DSBs, so it is not used in CRISPR-Cas9-mediated gene editing.

Why other options are incorrect:

A. NHEJ, B. MMEJ, D. SSA: All are DSB repair pathways actively exploited in CRISPR gene editing.

QUESTION

Arrange the following steps of base excision repair in order:

1. Endonuclease excises the sugar backbone
2. Glycosylase removes the altered base
3. DNA-polymerase adds a nucleotide
4. Creation of apurinic/apyrimidinic site

A 1 -> 2 -> 3 -> 4

B 2 -> 1 -> 4 -> 3

C 1 -> 2 -> 4 -> 3

D 2 -> 4 -> 1 -> 3

CORRECT

RIGHT ANSWER

OK D. 2 -> 4 -> 1 -> 3

WHY THIS IS RIGHT

Base excision repair (BER) proceeds in the following order:

1. Glycosylase removes the altered base (2): Recognizes and excises the damaged base, leaving an apurinic/apyrimidinic (AP) site.
2. Creation of AP site (4): The sugar-phosphate backbone remains intact, now missing the base.
3. Endonuclease excises the sugar backbone (1): AP endonuclease cleaves the phosphodiester bond at the AP site.
4. DNA polymerase adds a nucleotide (3): Fills in the gap using the complementary strand as a template.

Why other options are incorrect:

- A. 1 -> 2 -> 3 -> 4: Backbone is not excised first; base removal occurs first.
- B. 2 -> 1 -> 4 -> 3: AP site forms after base removal, before endonuclease action.
- C. 1 -> 2 -> 4 -> 3: Same issue-backbone excision occurs too early.

QUESTION

A mutation in a non-coding DNA sequence is believed to affect expression of the gene coding for a specific fetal enzyme. Liver and bone marrow cells from the fetus and his parents are obtained. Which of the following is the best method to determine if this gene is being transcribed in cultures of the isolated cells?

A Northern blot

CORRECT

B Western blot

C Southern blot

D Enzyme-linked immunosorbent assay

RIGHT ANSWER

OK A. Northern blot

WHY THIS IS RIGHT

Northern blotting detects specific RNA transcripts, allowing measurement of gene expression at the transcriptional level. In this case, it can determine whether the fetal enzyme gene is being transcribed in liver or bone marrow cells. It distinguishes mRNA presence, size, and relative abundance.

Why other options are incorrect:

- B. Western blot: Detects protein, not transcription; post-transcriptional or translational changes could confound results.
- C. Southern blot: Detects DNA sequences, used for identifying gene presence or mutations, not transcription.
- D. Enzyme-linked immunosorbent assay (ELISA): Measures protein concentration, not RNA expression.

QUESTION

Which of the following is not associated with post-transcriptional modification?

- A Methylation
- B Endonuclease cleavage
- C Adenylation

D Glycosylation

CORRECT

RIGHT ANSWER

D. Glycosylation

WHY THIS IS RIGHT

Glycosylation is a post-translational modification, not post-transcriptional. It occurs on proteins in the endoplasmic reticulum or Golgi apparatus after translation, adding carbohydrate moieties to amino acid residues.

Why other options are incorrect:

- A. Methylation: Can occur on RNA bases (e.g., 5' cap methylation), a post-transcriptional modification.
- B. Endonuclease cleavage: Involves splicing of pre-mRNA, a key post-transcriptional step.
- C. Adenylation: Addition of the poly-A tail at the 3' end of mRNA, also post-transcriptional.

QUESTION

| Which of the following does not happen in 5' to 3' direction?

A Transcription

B DNA replication

C DNA repair

D RNA editing

CORRECT

RIGHT ANSWER

OK D. RNA editing

WHY THIS IS RIGHT

RNA editing refers to post-transcriptional modification of RNA bases (e.g., A->I or C->U changes) and does not involve directional chain synthesis. Therefore, it does not proceed in a defined 5'→3' direction like nucleic acid synthesis processes.

Why other options are incorrect:

A. Transcription: RNA is synthesized 5'→3' by RNA polymerase.

B. DNA replication: DNA polymerases add nucleotides only in the 5'→3' direction.

C. DNA repair: Although excision occurs first, the resynthesis step by DNA polymerase still proceeds 5'→3'.

QUESTION

| Which gene is not involved in double strand DNA break repair mechanism?

A MRE11

B BRCA1

C KU80

D MuTL

CORRECT

RIGHT ANSWER

OK D. MuTL

WHY THIS IS RIGHT

Double-strand DNA break (DSB) repair is mediated by homologous recombination (HR) and non-homologous end joining (NHEJ) pathways. MRE11 is part of the MRN complex involved in sensing and processing DSBs. BRCA1 plays a crucial role in homologous recombination repair by facilitating end resection. KU80 is a key component of the NHEJ pathway, binding DNA ends. MutL, however, is involved in DNA mismatch repair, not in double-strand break repair.

Why other options are incorrect:

- A. MRE11: Essential for DSB recognition and end processing.
- B. BRCA1: Central to homologous recombination repair.
- C. KU80: Required for non-homologous end joining.

QUESTION

In which of the genetic material, Number of Adenine will not be equal to that of Thymine?

- A Pseudomonas
- B Mitochondrial DNA of human
- C Mycoplasma

D Measles

CORRECT

RIGHT ANSWER

OK D. Measles

WHY THIS IS RIGHT

Measles virus contains single-stranded RNA as its genetic material. Chargaff's rule ($A = T$, $G = C$) applies only to double-stranded DNA, where complementary base pairing enforces equality. In RNA viruses, thymine is absent (uracil replaces it), and base pairing rules do not mandate equal base proportions, so adenine will not equal thymine.

Why other options are incorrect:

- A. Pseudomonas: A bacterium with double-stranded DNA; follows Chargaff's rule.
- B. Mitochondrial DNA of humans: Though circular and bacterial-like, it is double-stranded DNA, so $A = T$.
- C. Mycoplasma: A bacterium with double-stranded DNA, despite small genome size; still obeys Chargaff's rule.

QUESTION

Which of the following is correct about monoclonal antibody production by hybridoma technique?

1. Antibody-producing B-lymphocytes are fused with immortal myeloma cell lines from mice
2. HAT medium contains Hypoxanthine-Aminopterin-Thymidine
3. HAT medium inhibits conversion of T→U
4. HGPRT is deficient in myeloma cells

A 1, 2, 3, 4

B 1, 2, 4

CORRECT

C 2, 3

D 1, 3, 4

RIGHT ANSWER

OK B. 1, 2, 4

WHY THIS IS RIGHT

In the hybridoma technique, antibody-producing B lymphocytes from an immunized mouse are fused with immortal myeloma cells to form hybridomas (Statement 1 true). Selection is done using HAT medium (Hypoxanthine-Aminopterin-Thymidine) (Statement 2 true). Myeloma cells are HGPRT-deficient, so they cannot survive in HAT medium unless fused with B cells (Statement 4 true).

Why other options are incorrect:

Statement 3: Aminopterin blocks de novo purine synthesis, not conversion of T → U.

Options including statement 3 are therefore incorrect.

QUESTION

Which of the following is true about DNA polymerase I?

A It is involved in the creation of Okazaki fragments.

B It is found in Eukaryotes

C It removes RNA primers.

CORRECT

D It is the primary enzyme of DNA synthesis.

RIGHT ANSWER

OK **C. It removes RNA primers.**

WHY THIS IS RIGHT

DNA polymerase I is a prokaryotic enzyme with unique 5'→3' exonuclease activity, which allows it to remove RNA primers from Okazaki fragments and replace them with DNA. It also has 3'→5' exonuclease activity for proofreading but is not the main enzyme for DNA elongation.

Why other options are incorrect:

A. Creation of Okazaki fragments: Done by primase and extended by DNA polymerase III, not polymerase I.

B. Found in eukaryotes: DNA polymerase I is exclusive to prokaryotes.

D. Primary enzyme of DNA synthesis: DNA polymerase III is the main replicative enzyme in prokaryotes.

QUESTION

Which of the following statements is incorrect about histones?

- A** They are large, positively charged molecules.
- B** They are made up primarily of arginine and lysine.
- C** They are produced during G2-M phase. CORRECT
- D** The H1 histone molecule is a linker molecule that is not a part of the nucleosome.

RIGHT ANSWER

- OK** **C. They are produced during G2-M phase.**

WHY THIS IS RIGHT

Histones are synthesized mainly during the S phase of the cell cycle to package newly replicated DNA into chromatin. Their production is tightly coupled with DNA replication to ensure proper nucleosome assembly. Hence, stating that histones are produced during the G2-M phase is incorrect.

Why other options are incorrect:

Large, positively charged molecules: True; positive charge allows binding to negatively charged DNA.

Made up primarily of arginine and lysine True; these basic amino acids confer positive charge.

H1 histone molecule is a linker that is not a part of nucleosome: True; H1 stabilizes DNA between nucleosomes and is not part of the core octamer.

QUESTION

Arrange the following disorders with the genes related to it:

1. Alzheimer's disease	A. ABCA2
2. Pulmonary alveolar proteinosis	B. ABCA3
3. Dubin Johnson syndrome	C. ABCC2
4. Tangier's disease	D. ABCA1

A 1-A, 2-B, 3-C, 4-D

CORRECT

B 1-B, 2-A, 3-C, 4-D

C 1-C, 2-B, 3-A, 4-D

D 1-A, 2-C, 3-B, 4-D

RIGHT ANSWER

OK A. 1-A, 2-B, 3-C, 4-D

WHY THIS IS RIGHT

1. Alzheimer's disease -> A. ABCA2: ABCA2 is expressed in brain tissue and is implicated in amyloid processing and neuronal lipid transport, linking it to Alzheimer's disease.
2. Pulmonary alveolar proteinosis -> B. ABCA3: ABCA3 is essential for surfactant transport in type II pneumocytes; mutation causes alveolar proteinosis.
3. Dubin-Johnson syndrome -> C. ABCC2: ABCC2 (MRP2) mediates bilirubin excretion into bile; mutation leads to conjugated hyperbilirubinemia.
4. Tangier disease -> D. ABCA1: ABCA1 is required for cholesterol efflux to HDL; deficiency causes Tangier disease.

Why other options are incorrect:

They mismatch transporter functions with unrelated organ systems (e.g., ABCA3 is lung-specific, ABCA1 is lipid/HDL related), leading to incorrect disease associations.

QUESTION

Chromosome painting is particularly useful for:

- A** Detecting point mutations in oncogenes
- B** Identifying cryptic translocations and complex chromosomal rearrangements CORRECT
- C** Measuring telomerase activity
- D** Quantifying gene expression levels

RIGHT ANSWER

- OK** **B. Identifying cryptic translocations and complex chromosomal rearrangements**

WHY THIS IS RIGHT

Chromosome painting uses fluorescent in situ hybridization (FISH) with chromosome-specific probes to visualize entire chromosomes. It is particularly useful for detecting cryptic translocations, marker chromosomes, and complex structural rearrangements that may be missed on routine karyotyping, especially in cancers and congenital abnormalities.

Why other options are incorrect:

- A. Detecting point mutations in oncogenes: Requires sequencing techniques, not chromosome painting.
- C. Measuring telomerase activity: Assessed by TRAP assay or molecular methods.
- D. Quantifying gene expression levels: Performed using RT-PCR or microarray/RNA-seq techniques.

QUESTION

DICER1 mutation-associated tumor syndrome commonly includes all of the following except:

- A** Wilm's tumor
- B** Sertoli-Leydig cell tumor
- C** Multinodular goiter

D Retinoblastoma

CORRECT

RIGHT ANSWER

OK D. Retinoblastoma

WHY THIS IS RIGHT

DICER1 mutation-associated tumor syndrome predisposes to tumors related to abnormal microRNA processing. Common manifestations include Wilms tumor, Sertoli-Leydig cell tumors of the ovary, and multinodular goiter. Retinoblastoma, however, is caused by mutations in the RB1 tumor suppressor gene and is not associated with DICER1 mutations.

Why other options are incorrect:

- A. Wilms tumor: Well-recognized association with DICER1 syndrome.
- B. Sertoli-Leydig cell tumor: Characteristic ovarian tumor in DICER1 mutation carriers.
- C. Multinodular goiter: Common endocrine manifestation of DICER1 syndrome.

QUESTION

Which of the following is false regarding eukaryotic initiation?

- A** Kozak sequence assists ribosomal scanning for AUG.
- B** eIF4A helicase unwinds secondary RNA structures.
- C** 43S complex interacts with 5' cap through eIF4E.
- D** **Formyl-methionine initiates protein synthesis**

CORRECT

RIGHT ANSWER

- OK** **D. Formyl-methionine initiates protein synthesis**

WHY THIS IS RIGHT

In eukaryotic initiation, translation begins with methionine, not formyl-methionine (fMet). fMet is a characteristic feature of prokaryotic translation, where it is used to initiate protein synthesis on bacterial ribosomes. Eukaryotes rely on a cap-dependent scanning mechanism and multiple initiation factors, without formylation of the initiator methionine.

Why other options are incorrect:

- A. Kozak sequence assists ribosomal scanning for AUG: True; it helps identify the AUG start codon.
- B. eIF4A helicase unwinds secondary RNA structures: True; it removes secondary structures in 5' UTR.
- C. 43S complex interacts with 5' cap via eIF4E: True; eIF4E binds the cap to recruit ribosomes.

QUESTION

A gene-rich region with repeated CG dinucleotide sequences adopts an alternating purine-pyrimidine structure. This form of DNA is often associated with transcriptional regulation and is stabilized during negative supercoiling. Which DNA form is this?

A A-DNA

B B-DNA

C Z-DNA

CORRECT

D C-DNA

RIGHT ANSWER

OK C. Z-DNA

WHY THIS IS RIGHT

Z-DNA is a left-handed DNA helix formed in gene-rich regions containing alternating purine-pyrimidine sequences, classically CG repeats. It is stabilized by negative supercoiling and is often found near actively transcribed genes, where it plays a role in transcriptional regulation and chromatin remodeling. Its zig-zag sugar-phosphate backbone gives it the characteristic “Z” shape.

Why other options are incorrect:

A. A-DNA: Right-handed helix seen in dehydrated DNA and RNA-DNA hybrids.

B. B-DNA: Normal right-handed DNA form, lacks alternating CG zig-zag structure.

D. C-DNA: Not a biologically relevant or well-defined DNA conformation.

QUESTION

A bacterial culture produces β -galactosidase even in absence of lactose. Which mutation is most likely?

- A** Mutation in lacZ
- B** Mutation in lacI CORRECT
- C** Mutation in promoter region reducing RNA polymerase binding
- D** Mutation in CAP binding site preventing cAMP activation

RIGHT ANSWER

- OK** **B. Mutation in lacI**

WHY THIS IS RIGHT

The lacI gene encodes the lac repressor, which normally binds the operator and prevents transcription of lac operon genes in the absence of lactose. A mutation in lacI produces a nonfunctional repressor that cannot bind the operator, resulting in constitutive expression of β -galactosidase, even without lactose.

Why other options are incorrect:

- A. Mutation in lacZ: Would reduce or abolish β -galactosidase production, not increase it.
- C. Mutation in promoter region: Decreased RNA polymerase binding would reduce transcription.
- D. Mutation in CAP binding site: Prevents cAMP-CAP activation, lowering transcription, especially in low glucose.

QUESTION

A 21-year-old female is evaluated for progressive gait instability and slurred speech. DNA analysis shows GAA repeat expansion causing decreased expression of a mitochondrial protein due to heterochromatin formation. What is the underlying mechanism?

A Anticipation due to gain-of-function in a cytoplasmic protein

B Transcriptional repression due to gene silencing

CORRECT

C mRNA instability due to nonsense-mediated decay

D Toxic RNA aggregation in neuronal nuclei

RIGHT ANSWER

OK **B. Transcriptional repression due to gene silencing**

WHY THIS IS RIGHT

This describes Friedreich ataxia, caused by GAA trinucleotide repeat expansion in the *FXN* gene. The expanded repeats induce heterochromatin formation, leading to epigenetic gene silencing and reduced transcription of frataxin, a mitochondrial protein involved in iron-sulfur cluster formation. The disease results from loss of function due to decreased gene expression, not from a toxic protein or RNA effect.

Why other options are incorrect:

A. Anticipation due to gain-of-function in a cytoplasmic protein: Friedreich ataxia is a loss-of-function disorder, not gain-of-function.

C. mRNA instability due to nonsense-mediated decay: There is no premature stop codon causing mRNA degradation.

D. Toxic RNA aggregation in neuronal nuclei: Toxic RNA aggregation occurs in disorders like myotonic dystrophy, not here.

QUESTION

The 5' cap of eukaryotic mRNA consists of:

- A** 3-methyladenosine
- B** 7-methylguanosine linked via 5'→5' triphosphate bond CORRECT
- C** 7-methyladenosine linked via 3'→5' bond
- D** 3-methyluridine

RIGHT ANSWER

- OK** **B. 7-methylguanosine linked via 5'→5' triphosphate bond**

WHY THIS IS RIGHT

The 5' cap of eukaryotic mRNA is formed by addition of 7-methylguanosine to the 5' end of the transcript through an unusual 5'→5' triphosphate linkage. This cap protects mRNA from exonuclease degradation, facilitates nuclear export, and is essential for ribosome binding and translation initiation via cap-binding proteins.

Why other options are incorrect:

- A. 3-methyladenosine: 3-methyladenosine is not a component of the mRNA cap.
- C. 7-methyladenosine linked via 3'→5' bond: The cap is guanosine, not adenosine, and linkage is not 3'→5'.
- D. 3-methyluridine: 3-methyluridine is unrelated to mRNA capping.

QUESTION

A mutation from GCCACCATGG to GCCACCCTGG leads to decreased protein translation without affecting mRNA level. Which step is impaired?

A RNA polymerase binding

B Ribosome scanning and start codon recognition

CORRECT

C 5' capping

D Polyadenylation

RIGHT ANSWER

OK **B. Ribosome scanning and start codon recognition**

WHY THIS IS RIGHT

(Kozak sequence -> important for AUG recognition)

The sequence GCCACCATGG represents the Kozak consensus sequence, which is critical for efficient ribosomal recognition of the AUG start codon during translation initiation. The mutation to GCCACCCTGG disrupts the AUG start codon context, impairing ribosome scanning and start codon recognition. As a result, translation efficiency decreases, even though mRNA levels remain normal, indicating a post-transcriptional defect.

Why other options are incorrect:

- A. RNA polymerase binding: Would affect transcription and mRNA levels, not translation alone.
- C. 5' capping: Required for mRNA stability and ribosome binding; defects reduce mRNA levels.
- D. Polyadenylation: Affects mRNA stability and nuclear export, also reducing mRNA levels

QUESTION

| Which of the following is an example of facilitated diffusion across a cell membrane?

A Glucose transport via GLUT4

CORRECT

B Sodium-potassium pump

C Endocytosis

D Water movement through aquaporins

RIGHT ANSWER

OK

A. Glucose transport via GLUT4

WHY THIS IS RIGHT

Facilitated diffusion is a type of passive transport in which molecules move down their concentration gradient across the cell membrane with the help of specific carrier or channel proteins, without the use of ATP.

Glucose transport via GLUT4 is a classic example of facilitated diffusion. GLUT4 is an insulin-regulated glucose transporter found mainly in skeletal muscle and adipose tissue. After insulin stimulation, GLUT4 transporters move to the cell membrane and allow glucose to enter the cell along its concentration gradient without energy expenditure.

Why other options are incorrect:

- Sodium-potassium pump: This is primary active transport that uses ATP to move Na⁺ out and K⁺ into the cell against their gradients.
- Endocytosis: A vesicular transport mechanism that requires energy and is not diffusion.
- Water movement through aquaporins: This is osmosis through channel proteins, not carrier-mediated facilitated diffusion.

QUESTION

Mechanism of action of botulinum toxin is:

- A** Inhibits NM receptor
- B** Inhibits reuptake of acetyl choline
- C** **Inhibits release of acetylcholine from lower motor neurons**
- D** Inhibits synthesis of acetyl choline

CORRECT

RIGHT ANSWER

- OK** **C. Inhibits release of acetylcholine from lower motor neurons**

WHY THIS IS RIGHT

Botulinum toxin blocks neuromuscular transmission by inhibiting the release of acetylcholine (ACh) at the presynaptic terminals of lower motor neurons.

Mechanism:

- Botulinum toxin cleaves SNARE proteins (e.g., synaptobrevin, SNAP-25).
- These proteins are essential for fusion of synaptic vesicles with the presynaptic membrane.
- Their inhibition prevents exocytosis of acetylcholine, leading to failure of neuromuscular transmission.

Clinical correlation:

- Causes descending flaccid paralysis (e.g., botulism)
- Used therapeutically in conditions like dystonia, spasticity, and cosmetic procedures

Why other options are incorrect:

- Inhibits NM receptor: Seen with drugs like curare, not botulinum toxin.
- Inhibits reuptake of acetylcholine: ACh is not reuptaken like monoamines; it is degraded by acetylcholinesterase.
- Inhibits synthesis of acetylcholine: Not the mechanism of botulinum toxin.

QUESTION

| Which of the following is seen in low insulin: glucagon ratio?

- A** Activation of lipoprotein lipase
- B** Activation of glycogen synthase
- C** Activation of phosphofructokinase 1

D Activation of hormone-sensitive lipase

CORRECT

RIGHT ANSWER

OK D. Activation of hormone-sensitive lipase

WHY THIS IS RIGHT

A low insulin: glucagon ratio represents the fasting/starvation state, where the body shifts from energy storage to energy mobilization.

Metabolic effects of low insulin-high glucagon state:

- ↑ Lipolysis in adipose tissue
- Activation of hormone-sensitive lipase (HSL) -> breakdown of triglycerides into free fatty acids and glycerol
- Fatty acids are used for β -oxidation and ketone body formation

Why other options are incorrect:

- Lipoprotein lipase: Activated by insulin (fed state) for fat storage
- Glycogen synthase: Active in fed state (high insulin) for glycogen formation
- Phosphofructokinase-1 (PFK-1): Activated in glycolysis (fed state), not during fasting

QUESTION

A diabetic patient was advised exercise by the treating physician. The glucose transporter which is insulin sensitive is:

A Glut 1

B Glut 2

C Glut 3

D Glut 4

CORRECT

RIGHT ANSWER

OK D. Glut 4

WHY THIS IS RIGHT

GLUT-4 is the insulin-sensitive glucose transporter, primarily found in:

- Skeletal muscle
- Adipose tissue

Mechanism:

- In response to insulin, GLUT-4 translocates from intracellular vesicles to the cell membrane
- This increases glucose uptake into cells
- Exercise can also stimulate GLUT-4 translocation independent of insulin

Clinical relevance in diabetes:

- Exercise improves glucose uptake even when insulin action is impaired
- Hence, exercise is beneficial in diabetes management

Why other options are incorrect:

- GLUT-1: RBCs, brain (insulin-independent)
- GLUT-2: Liver, pancreas (high K_m , bidirectional)
- GLUT-3: Neurons (high affinity, insulin-independent)

QUESTION

A miner has been trapped in a tunnel for the past 5 days without access to food. What is the primary source of energy for the brain under these conditions?

A Glycogenolysis

B Ketosis

CORRECT

C Gluconeogenesis

D Lipolysis

RIGHT ANSWER

OK B. Ketosis

WHY THIS IS RIGHT

During prolonged fasting/starvation ($\geq 3-5$ days), the body adapts to conserve glucose. The brain, which normally depends on glucose, shifts to using ketone bodies as its primary energy source.

Time Period	Main Energy Source	Brain Fuel
0-18 hrs	Hepatic glycogenolysis	Glucose
18-48 hrs	Gluconeogenesis (AA, glycerol, lactate)	Glucose
>48 hrs / 5 days	Ketogenesis	Ketone bodies

QUESTION

Entry of glucose is insulin independent in all of them except:

- A RBC
- B Pancreas
- C Adipose tissues CORRECT
- D Brain

RIGHT ANSWER

OK C. Adipose tissues

WHY THIS IS RIGHT

Major Glucose transporters and their function is summarized as:

Transporter	Location	Function
Glut-1	Brain, Kidney, Placenta, Erythrocytes, Cancer	Basal uptake of glucose, High affinity
Glut-2	Liver, Pancreatic cell, small intestine	Fed state
Glut-3	Brain, Kidney, Placenta	Glucose uptake, Maximum affinity for glucose
Glut-4	Heart, Skeletal muscle, Adipose tissue	Insulin-stimulated glucose uptake
Glut-5	Small intestine, Sperms	Fructose

QUESTION

| A patient diagnosed with diabetes has high increased LDL and VLDL because of ?

- A** Hormone sensitive lipase and lipoprotein lipase both increased
- B** **Hormone sensitive lipase increased & lipoprotein lipase decreased**
- C** Lipoprotein lipase increased & hormone sensitive lipase decreased
- D** Lipoprotein lipase and hormone sensitive lipase both decreased

CORRECT

RIGHT ANSWER

- OK** **B. Hormone sensitive lipase increased & lipoprotein lipase decreased**

WHY THIS IS RIGHT

In diabetes mellitus (especially uncontrolled diabetes), insulin deficiency or resistance alters lipid metabolism, leading to hypertriglyceridemia and increased VLDL and LDL levels.

1\ Effect on Hormone-Sensitive Lipase (HSL)

- Insulin normally inhibits HSL in adipose tissue.
- When insulin is deficient -> HSL activity increases.
- This causes enhanced lipolysis in adipose tissue, releasing large amounts of free fatty acids (FFA) into the circulation.
- The liver takes up these FFAs and converts them into triglycerides, which are packaged as VLDL -> increased VLDL production.

2\ Effect on Lipoprotein Lipase (LPL)

- Insulin normally stimulates LPL, which hydrolyzes triglycerides in VLDL and chylomicrons to allow uptake into tissues.
- In diabetes -> LPL activity decreases due to lack of insulin.
- This results in reduced clearance of VLDL and chylomicrons, further increasing plasma triglycerides.

QUESTION

Which of the following tissues is unable to take up glucose in the absence of insulin?

A Skeletal muscles

CORRECT

B RBC

C Brain

D Kidney

RIGHT ANSWER

OK A. Skeletal muscles

WHY THIS IS RIGHT

Glucose uptake in different tissues depends on specific glucose transporters (GLUTs), some of which are insulin dependent.

- Skeletal muscle and adipose tissue require insulin for glucose entry.
- Insulin stimulates translocation of GLUT-4 transporters to the cell membrane.
- Without insulin, GLUT-4 does not move to the membrane, so glucose uptake is markedly reduced.

Therefore, skeletal muscle cannot effectively take up glucose in the absence of insulin.

Some tissues use transporters that do not require insulin:

- RBCs -> GLUT-1
- Brain -> GLUT-1 and GLUT-3
- Kidney, liver, pancreatic β -cells -> GLUT-2

These tissues can take up glucose even during fasting or insulin deficiency.

QUESTION

In a 90 kg man following a meal, which of the following enzymes or intermediates is most likely activated to promote anabolic processes contributing to weight gain?

1. Malonyl CoA
2. Acetyl CoA carboxylase
3. Mitochondrial carnitine transporter
4. Tricarboxylate transporter (citrate)

A 1, 2 and 4

CORRECT

B 1 and 2

C 1 and 3

D 1, 2, 3 and 4

RIGHT ANSWER

OK A. 1, 2 and 4

WHY THIS IS RIGHT

In the fed state (post-meal), insulin promotes anabolic metabolism, especially fatty acid synthesis and storage, contributing to weight gain.

Key Activated Components:

- Acetyl-CoA carboxylase (ACC): Activated by insulin → converts acetyl-CoA to malonyl-CoA (rate-limiting step of fatty acid synthesis).
- Malonyl-CoA: Promotes lipogenesis and inhibits fatty acid oxidation.
- Tricarboxylate transporter (citrate shuttle): Moves citrate from mitochondria to cytosol to provide acetyl-CoA for fatty acid synthesis.

Malonyl-CoA inhibits the mitochondrial carnitine transporter (CPT-1), preventing fatty acid entry into mitochondria for β -oxidation.

QUESTION

An infant came with vomiting, jaundice, and hypoglycemia. What is the most likely diagnosis?

A Galactosemia

CORRECT

B Phenylketonuria

C Wilson disease

D Menke disease

RIGHT ANSWER

A. Galactosemia

WHY THIS IS RIGHT

Galactosemia is an inborn error of metabolism caused by deficiency of galactose-1-phosphate uridylyltransferase, leading to accumulation of toxic metabolites after milk ingestion.

- Presents in neonates/infants after starting milk feeds
- Features include vomiting, jaundice, hypoglycemia, hepatomegaly, and failure to thrive
- Can also cause cataracts and risk of E. coli sepsis
- Early diagnosis is crucial
- Management requires immediate elimination of galactose/lactose from diet

Contrast:

- Phenylketonuria: intellectual disability, musty odor, no jaundice/hypoglycemia
- Wilson disease: presents later with liver and neurological features
- Menkes disease: copper metabolism disorder with kinky hair and neurodegeneration

QUESTION

A 1-week-old child was presented with early cataract. He also has refusal to feed, vomiting, lethargy, jaundice, and sepsis. This can be caused due to the deficiency of which enzyme?

A Catalase

B Galactose-1-phosphate uridylyltransferase

CORRECT

C Aldose reductase

D Hexokinase

RIGHT ANSWER

OK

B. Galactose-1-phosphate uridylyltransferase

WHY THIS IS RIGHT

Classic galactosemia is caused by deficiency of galactose-1-phosphate uridylyltransferase (GALT) and presents in neonates soon after milk feeding.

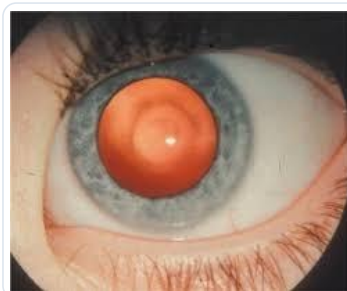
- Early cataract (due to galactitol accumulation in lens)
- Poor feeding, vomiting, lethargy
- Jaundice and hepatomegaly
- Failure to thrive
- Risk of E. coli sepsis

Biochemical basis:

- Block in conversion of galactose-1-phosphate → UDP-galactose
- Accumulation of galactose-1-phosphate (toxic) and galactitol

Why not others:

- Aldose reductase deficiency: Would reduce cataract formation (not cause disease)
- Hexokinase: General glycolysis enzyme, not specific
- Catalase: Involved in oxidative stress, unrelated



QUESTION

| Which of the following enzyme is deficient in Fabry's Disease?

CORRECT

A α -Galactosidase

B β -Galactosidase

C β -Glucosidase

D Sphingomyelinase

RIGHT ANSWER

OK **A. α -Galactosidase**

WHY THIS IS RIGHT

Fabry disease is a lysosomal storage disorder caused by deficiency of the enzyme α -galactosidase A. This enzyme normally breaks down globotriaosylceramide (ceramide trihexoside). When the enzyme is deficient, these glycosphingolipids accumulate in vascular endothelium, kidneys, heart, and nerves, leading to progressive organ damage.

Typical clinical features:

- Angiokeratomas (skin lesions)
- Acroparesthesia (burning pain in hands and feet)
- Hypohidrosis
- Progressive renal failure and cardiomyopathy

Why other options are incorrect:

- β -Galactosidase deficiency: Causes GM1 gangliosidosis.
- β -Glucosidase deficiency: Causes Gaucher disease.
- Sphingomyelinase deficiency: Causes Niemann-Pick disease.

QUESTION

A child presents with abdominal distension, fasting hypoglycemia, and hepatomegaly (liver palpable 8 cm below the costal margin). The liver is smooth on palpation. Which of the following enzyme deficiencies is the most likely cause?

A Glucose-6-phosphatase

CORRECT

B Glucose-6-phosphate dehydrogenase

C Myophosphorylase

D Phosphofructokinase

RIGHT ANSWER

OK

A. Glucose-6-phosphatase

WHY THIS IS RIGHT

Deficiency of glucose-6-phosphatase causes Glycogen Storage Disease Type I (Von Gierke disease). This enzyme normally converts glucose-6-phosphate to free glucose in the final step of glycogenolysis and gluconeogenesis in the liver and kidney.

When this enzyme is deficient:

- Glucose cannot be released into the bloodstream, leading to severe fasting hypoglycemia.
- Glucose-6-phosphate accumulates, which is diverted into glycogen synthesis, causing excess glycogen deposition in the liver.
- This results in marked hepatomegaly and abdominal distension.

Why other options are incorrect:

- Glucose-6-phosphate dehydrogenase deficiency: Causes hemolytic anemia due to impaired NADPH production in RBCs, not hepatomegaly with hypoglycemia.
- Myophosphorylase deficiency: Causes McArdle disease, presenting with exercise intolerance and muscle cramps, not liver enlargement.
- Phosphofructokinase deficiency: Causes Tarui disease (GSD VII) with muscle weakness and hemolysis.

QUESTION

A 2-week-old exclusively breastfed infant is brought with poor feeding, vomiting, lethargy, and failure to thrive. On examination, the infant has hepatomegaly and bilateral cataracts. Laboratory evaluation shows hypoglycemia and elevated liver enzymes. Which of the following enzyme deficiencies is most likely responsible for this presentation?

A Galactokinase

B Galactose-1-phosphate uridyl transferase (GALT)

CORRECT

C Aldolase B

D Fructokinase

RIGHT ANSWER

OK **B. Galactose-1-phosphate uridyl transferase (GALT)**

WHY THIS IS RIGHT

Classic galactosemia is caused by deficiency of galactose-1-phosphate uridyl transferase (GALT), an enzyme involved in the metabolism of galactose. This condition commonly presents in breastfed infants, because breast milk contains lactose, which is broken down into glucose and galactose.

When GALT is deficient, galactose-1-phosphate and galactose accumulate in tissues such as the liver, brain, and lens of the eye. Excess galactose is converted to galactitol, which accumulates in the lens and causes cataracts. Accumulation in the liver leads to hepatomegaly and liver dysfunction, while toxic metabolites contribute to lethargy and feeding difficulties.

Why other options are incorrect:

- Galactokinase deficiency: Causes cataracts only, without severe liver involvement or systemic toxicity.
- Aldolase B deficiency: Seen in hereditary fructose intolerance, presenting after fructose ingestion.
- Fructokinase deficiency: Causes essential fructosuria, a benign condition.

QUESTION

| Rice water diarrhoea, intoxication is due to which ganglioside?

CORRECT

A GM-1 ganglioside receptor

B GM-2 ganglioside receptor

C GM-3 ganglioside receptor

D GD-1a ganglioside receptor

RIGHT ANSWER

OK **A. GM-1 ganglioside receptor**

WHY THIS IS RIGHT

Rice water diarrhea" is characteristic of cholera, caused by *Vibrio cholerae*. The cholera toxin specifically binds to the GM1 ganglioside receptor present on the intestinal epithelial cell membrane.

- Cholera toxin binds to GM1 ganglioside on enterocytes.
- The toxin activates adenylate cyclase via Gs protein stimulation.
- This leads to ↑ cAMP levels.
- Increased cAMP causes excess secretion of chloride and water into the intestinal lumen.

Result:

- > Profuse watery diarrhea ("rice water stools")
- > Severe dehydration and electrolyte imbalance

Why other options are incorrect:

- GM2 ganglioside: Associated with Tay-Sachs disease metabolism, not toxin binding.
- GM3 ganglioside: Not involved in cholera toxin action.
- GD1a ganglioside: Involved in other neuronal interactions, not cholera.

QUESTION

A Mediterranean person didn't receive primaquine. Which pathway is affected by G-6-PD enzyme deficiency?

A HMP pathway

CORRECT

B Glycolysis

C Gluconeogenesis

D Kreb cycle

RIGHT ANSWER

OK A. HMP pathway

WHY THIS IS RIGHT

Glucose-6-phosphate dehydrogenase (G6PD) is the key regulatory enzyme of the Hexose Monophosphate (HMP) pathway / Pentose Phosphate Pathway.

This pathway generates NADPH, which is essential for:

- Maintaining reduced glutathione (GSH) in red blood cells
- Protecting RBCs from oxidative damage

In G6PD deficiency (common in Mediterranean populations):

- ↓ NADPH → ↓ reduced glutathione
- RBCs become vulnerable to oxidative stress (e.g., drugs like primaquine, infections, fava beans)
- Leads to hemolysis

Why other options are incorrect:

- Glycolysis: Generates ATP, not primarily dependent on G6PD
- Gluconeogenesis: Produces glucose, unrelated to G6PD
- Krebs cycle: Occurs in mitochondria, not affected by G6PD deficiency

QUESTION

| True about Warburg effect?

- A** Makes cancer cells immortal
- B** **Aerobic glycolysis with lactate formation**
- C** Produce more ATP
- D** Decrease glucose uptake

CORRECT

RIGHT ANSWER

- OK** **B. Aerobic glycolysis with lactate formation**

WHY THIS IS RIGHT

The Warburg effect refers to the preference of cancer cells to utilize aerobic glycolysis, meaning they convert glucose to lactate even in the presence of adequate oxygen.

Normally: Oxygen present -> cells use oxidative phosphorylation (high ATP yield)

In cancer cells: Despite oxygen -> glycolysis predominates -> lactate production

Why does this happen?

- Provides rapid ATP (though less efficient)
- Generates intermediates for biosynthesis (nucleotides, lipids, amino acids) needed for rapid cell proliferation

Why other options are incorrect:

- Makes cancer cells immortal: Immortality is due to telomerase activation, not Warburg effect
- Produce more ATP: Glycolysis produces less ATP than oxidative phosphorylation
- Decrease glucose uptake: Cancer cells actually show increased glucose uptake (basis of PET scan)

QUESTION

Cherry red spots on retina and mental retardation is seen in:



A Gaucher's disease

B **Tay Sach's disease**

CORRECT

C Hunter syndrome

D Krabbe' disease

RIGHT ANSWER

OK **B. Tay Sach's disease**

WHY THIS IS RIGHT

A cherry red spot on the retina with neurodegeneration (mental retardation) is characteristic of Tay-Sachs disease, a lysosomal storage disorder caused by deficiency of hexosaminidase A.

- Deficiency of hexosaminidase A -> accumulation of GM2 ganglioside in neurons
- Leads to progressive neurodegeneration
- The surrounding retina becomes pale due to lipid accumulation, while the fovea appears bright red (cherry red spot) because it lacks ganglion cells

Important distinguishing feature:

- No hepatosplenomegaly (helps differentiate from Niemann-Pick disease, which also has cherry red spot but with organomegaly)

Why other options are incorrect:

- Gaucher disease: Hepatosplenomegaly, bone crises (no cherry red spot)
- Hunter syndrome: Mucopolysaccharidosis with coarse facies, no cherry red spot
- Krabbe disease: Demyelination with globoid cells, no cherry red spot

QUESTION

A 16-year-old boy presents with exercise intolerance, early fatigue, and painful muscle cramps during strenuous activity. He reports that symptoms improve with rest. There is no significant rise in serum lactate levels after exercise testing. Serum creatine kinase is mildly elevated. Which of the following enzyme deficiencies best explains this condition?

A Myophosphorylase

CORRECT

B Glucose-6-phosphatase

C Glycogen phosphorylase (hepatic)

D Hexokinase

RIGHT ANSWER

OK **A. Myophosphorylase**

WHY THIS IS RIGHT

McArdle disease (Glycogen Storage Disease Type V) is caused by deficiency of myophosphorylase, the muscle isoform of glycogen phosphorylase, which is required for breakdown of glycogen to glucose-1-phosphate in skeletal muscle.

- Inability to utilize muscle glycogen during exercise
- Leads to energy deficiency in muscle cells during exertion

Clinical features:

- Exercise intolerance (early fatigue)
- Muscle cramps and pain
- Myoglobinuria (dark urine after exercise)
- "Second-wind phenomenon" (symptoms improve after rest due to use of blood glucose)

Why other options are incorrect:

- Glucose-6-phosphatase: Deficient in Von Gierke disease (GSD I)
- Glycogen phosphorylase: General term; the specific deficient enzyme in muscle is myophosphorylase
- Hexokinase: Involved in glycolysis, not glycogen breakdown

QUESTION

Which of the following acts as an inhibitor of Complex IV (Cytochrome c oxidase) in the electron transport chain?

A Cyanide

CORRECT

B Carbon Dioxide

C Oligomycin

D Antimycin

RIGHT ANSWER

OK

A. Cyanide

WHY THIS IS RIGHT

Complex IV (Cytochrome c oxidase) is the final enzyme of the electron transport chain (ETC) and is responsible for transferring electrons to oxygen, forming water.

Cyanide is a potent inhibitor of Complex IV. It binds to the ferric (Fe^{3+}) iron of cytochrome a_3 , blocking electron transfer to oxygen. This halts the entire ETC, leading to:

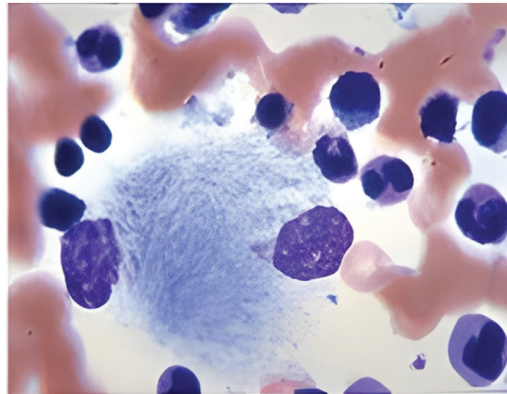
- Cessation of oxidative phosphorylation
- ↓ ATP production
- Cells switch to anaerobic glycolysis → lactic acidosis
- Causes histotoxic hypoxia (oxygen is present but cannot be utilized)

Why other options are incorrect:

- Carbon dioxide: Not an ETC inhibitor
- Oligomycin: Inhibits ATP synthase (Complex V)
- Antimycin A: Inhibits Complex III

QUESTION

The following histological image is seen in the patient who is complaining of growth failure and abdominal distension. Signs found were anemia, thrombocytopenia and bony pain. Which enzyme is deficient in this patient?



CORRECT

A Beta-Glucocerebrosidase

B Sphingomyelinase

C Hexosaminidase

D G-6-phosphatase

RIGHT ANSWER

A. Beta-Glucocerebrosidase

WHY THIS IS RIGHT

The histological image shows a Gaucher cell—a lipid-laden macrophage with a “crumpled tissue paper” cytoplasm, which is characteristic of Gaucher disease.

- Deficiency of β -glucocerebrosidase (β -glucosidase)
- Leads to accumulation of glucocerebroside in macrophages of the liver, spleen, and bone marrow

Clinical features explained:

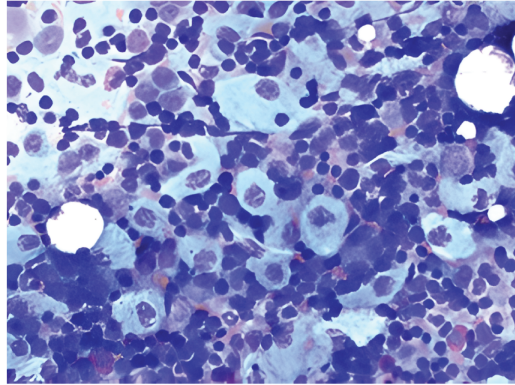
- Hepatosplenomegaly → abdominal distension
- Bone marrow infiltration → anemia + thrombocytopenia
- Bone involvement → bone pain and fractures
- Growth failure in children

Why other options are incorrect:

- Sphingomyelinase deficiency: Niemann-Pick disease (foam cells, neurodegeneration)
- Hexosaminidase deficiency: Tay-Sachs disease (cherry red spot, no organomegaly)
- G-6-phosphatase deficiency: Von Gierke disease (hypoglycemia, hepatomegaly)

QUESTION

An individual undergoes a bone marrow examination which shows the presence of the cells as shown in the image. Which of the following is the likely enzyme defect associated with this condition?



A Hexosaminidase

B Glucocerebrosidase

CORRECT

C Alpha 1,4 glucosidase

D Galactosidase

RIGHT ANSWER

B. Glucocerebrosidase

WHY THIS IS RIGHT

The bone marrow image shows Gaucher cells-large macrophages with abundant, wrinkled (“crumpled tissue paper”) cytoplasm, which is characteristic of Gaucher disease.

- Deficiency of glucocerebrosidase (β -glucosidase)
- Leads to accumulation of glucocerebroside in macrophages

Pathophysiology:

- Lipid-laden macrophages infiltrate bone marrow, liver, and spleen
- Causes marrow suppression $\rightarrow$ anemia, thrombocytopenia
- Bone involvement $\rightarrow$ bone pain, fractures
- Hepatosplenomegaly

Why other options are incorrect:

- Hexosaminidase: Tay-Sachs disease (neurodegeneration, cherry red spot)
- Alpha-1,4-glucosidase: Pompe disease (cardiomegaly, muscle weakness)
- Galactosidase: Fabry disease (angiokeratomas, neuropathy)

QUESTION

A child presents with mental retardation, bone pain, and inability to walk. On fundoscopy examination, a cherry red spot is seen. There is no organomegaly. What is the most likely diagnosis?

A Tay-Sachs disease

CORRECT

B Niemann-Pick disease

C Hurler syndrome

D Gaucher disease

RIGHT ANSWER

OK **A. Tay-Sachs disease**

WHY THIS IS RIGHT

A child with neurodegeneration (mental retardation), motor difficulty, and cherry red spot on fundoscopy without organomegaly is characteristic of Tay-Sachs disease.

- Deficiency of hexosaminidase A
- Leads to accumulation of GM2 ganglioside in neurons
- Lipid accumulation in neurons -> progressive neurodegeneration
- Retina appears pale due to storage -> fovea stands out as cherry red spot

Important differentiation:

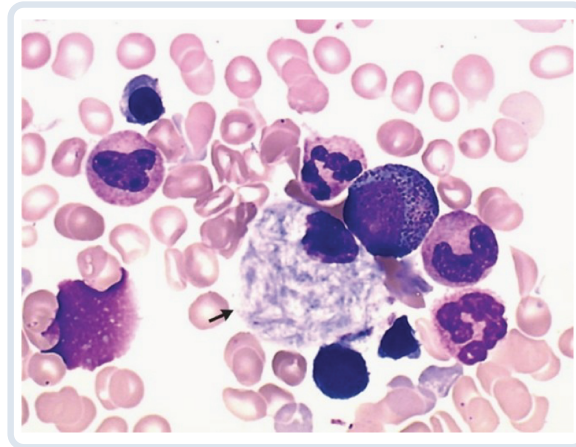
- Tay-Sachs: Cherry red spot without organomegaly
- Niemann-Pick: Cherry red spot with hepatosplenomegaly

Why other options are incorrect:

- Niemann-Pick disease: Has hepatosplenomegaly (foam cells)
- Hurler syndrome: Coarse facies, corneal clouding (no cherry red spot)
- Gaucher disease: Hepatosplenomegaly, bone pain, no cherry red spot

QUESTION

A child presents with bone pain and hepatosplenomegaly. A trephine biopsy and aspirate show the following finding. Which of the following is the most likely enzyme deficient in this condition?



A Hexosaminidase A

B. Glucocerebrosidase

CORRECT

C Sphingomyelinase

D α -1,4-Glucosidase

RIGHT ANSWER

B. Glucocerebrosidase

WHY THIS IS RIGHT

A child with bone pain, hepatosplenomegaly, and characteristic bone marrow findings (Gaucher cells) is diagnosed with Gaucher disease, a lysosomal storage disorder.

Underlying enzyme defect: Deficiency of glucocerebrosidase (β -glucosidase)

- Accumulation of glucocerebroside in macrophages
- These macrophages appear as Gaucher cells with "crumpled tissue paper" cytoplasm
- Infiltration of organs leads to:

- o Hepatosplenomegaly
- o Bone marrow suppression -> anemia, thrombocytopenia
- o Bone pain and fractures

Why other options are incorrect:

- Hexosaminidase A: Tay-Sachs (cherry red spot, no organomegaly)
- Sphingomyelinase: Niemann-Pick (foam cells + hepatosplenomegaly)
- α -1,4-glucosidase: Pompe disease (cardiomegaly, muscle weakness)

QUESTION

A chronic alcoholic patient is diagnosed with gouty arthritis. Mark the correct biochemical changes which will be seen in this patient?

- A** Decreased NADH/NAD⁺ ratio
- B** Increased urea and urate levels
- C** Increased urate and lactate levels
- D** Increased level of alkaline phosphatase

CORRECT

RIGHT ANSWER

- OK** **C. Increased urate and lactate levels**

WHY THIS IS RIGHT

In chronic alcoholism, metabolism of ethanol in the liver leads to an increased NADH/NAD⁺ ratio due to the actions of alcohol dehydrogenase and aldehyde dehydrogenase.

Metabolic consequences:

- ↑ NADH shifts pyruvate → lactate → causes lactic acidosis
- Lactate competes with uric acid for renal excretion
- ↓ Excretion of uric acid → hyperuricemia → gout

Why other options are incorrect:

- Decreased NADH/NAD⁺ ratio: Opposite; alcohol increases NADH
- Increased urea and urate: Urea is not the key change linked to gout here
- Increased alkaline phosphatase: Not specific to gout or alcohol metabolism

QUESTION

A 4-year-old child presents with easy fatiguability. The mother also complains that the child has increased hunger between meals, which is relieved after food. Liver examination revealed no glycogen. What enzyme is most likely deficient?

A Glycogen phosphorylase

B Glycogen synthase

CORRECT

C Debranching enzyme

D Glucose 6 phosphatase

RIGHT ANSWER

OK

B. Glycogen synthase

WHY THIS IS RIGHT

Glycogen synthase deficiency (Glycogen Storage Disease Type 0) is characterized by an inability to synthesize glycogen, especially in the liver.

- Glycogen synthase catalyzes formation of glycogen from glucose
- Deficiency -> absent/markedly reduced glycogen stores in liver

Clinical features explained:

- Fasting hypoglycemia -> easy fatigability
- Increased hunger between meals (ketotic hypoglycemia)
- Symptoms improve after feeding
- No hepatomegaly (because glycogen is not stored)

Why other options are incorrect:

- Glycogen phosphorylase: Deficiency causes Hers disease (GSD VI) -> hepatomegaly
- Debranching enzyme: Causes Cori disease (GSD III) -> hepatomegaly + abnormal glycogen
- Glucose-6-phosphatase: Causes Von Gierke disease (GSD I) -> severe hypoglycemia + hepatomegaly

QUESTION

A 12-year-old boy is brought to the OPD as he is always fatigued and has muscle cramps after exercise and the blood sample collected soon after exercise showed low glucose and lactate level. He also has a history of one episode of hypoglycaemia while playing. He became normal after resting for a while. Which of the following diseases is he most likely to be suffering from?

A McArdle disease

CORRECT

B Hers disease

C Cori's disease

D Andersen disease

RIGHT ANSWER

OK **A. McArdle disease**

WHY THIS IS RIGHT

McArdle disease (Glycogen Storage Disease Type V) is caused by deficiency of myophosphorylase, the enzyme responsible for breakdown of glycogen in skeletal muscle.

- Muscle cannot convert glycogen → glucose-1-phosphate
- Leads to failure of glycolysis during exercise
- Hence, ↓ lactate production after exercise (key diagnostic clue)

Clinical features explained:

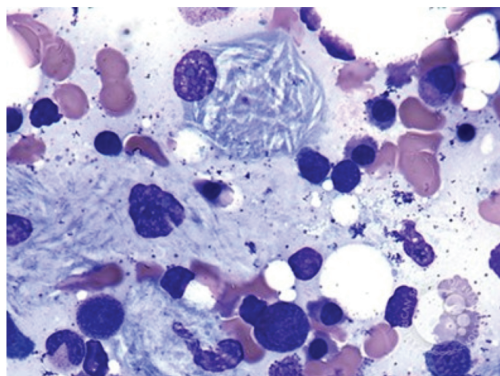
- Exercise intolerance & early fatigue
- Muscle cramps after exercise
- Low lactate levels post-exercise
- Second-wind phenomenon (symptoms improve after rest due to use of blood glucose)
- Occasional hypoglycemia during exertion

Why other options are incorrect:

- Hers disease (GSD VI): Liver glycogen phosphorylase deficiency → mild hypoglycemia + hepatomegaly
- Cori disease (GSD III): Debranching enzyme deficiency → hepatomegaly + abnormal glycogen
- Andersen disease (GSD IV): Branching enzyme deficiency → liver cirrhosis, not exercise intolerance

QUESTION

A crumpled tissue paper appearance and pancytopenia are seen in which enzyme defect?



A Alpha galactosidase

B Alpha glucosidase

C Beta galactosidase

D Glucocerebrosidase

CORRECT

RIGHT ANSWER

OK

D. Glucocerebrosidase

WHY THIS IS RIGHT

The “crumpled tissue paper” appearance of macrophages in bone marrow is characteristic of Gaucher disease, caused by deficiency of glucocerebrosidase.

- Deficiency of β -glucocerebrosidase
- Leads to accumulation of glucocerebroside in macrophages
- Lipid-laden macrophages (Gaucher cells) accumulate in:

- o Bone marrow
- o Liver
- o Spleen
- Causes pancytopenia due to marrow infiltration

Clinical features:

- Hepatosplenomegaly
- Pancytopenia (anemia, thrombocytopenia)
- Bone pain and fractures

Why other options are incorrect:

- Alpha-galactosidase: Fabry disease
- Alpha-glucosidase: Pompe disease
- Beta-galactosidase: GM1 gangliosidosis

QUESTION

A neonate was brought with complaints of poor feeding, vomiting, acidosis, and cataract. Urine Benedict's test was positive, while urine glucose was negative. The enzyme defective in this case is:

A Fructokinase

B Sucrase

C Galactose-1-phosphate uridyl transferase

CORRECT

D Lactase

RIGHT ANSWER

OK C. Galactose-1-phosphate uridyl transferase

WHY THIS IS RIGHT

A neonate with poor feeding, vomiting, acidosis, cataracts, and positive Benedict's test (reducing sugar present) but negative glucose is classic for galactosemia.

Deficiency of galactose-1-phosphate uridyl transferase (GALT)

- Inability to convert galactose-1-phosphate → UDP-galactose
- Accumulation of:

o Galactose-1-phosphate (toxic to liver)

o Galactitol (via aldose reductase → causes cataracts)

Clinical features explained:

- Vomiting, poor feeding: After milk intake (lactose → glucose + galactose)
- Hepatic dysfunction → acidosis
- Cataracts: Due to galactitol accumulation
- Benedict's positive, glucose negative: Indicates non-glucose reducing sugar (galactose)

Why other options are incorrect:

- Fructokinase: Essential fructosuria (benign)
- Sucrase: Causes sucrose intolerance (no cataracts)
- Lactase: Causes lactose intolerance (diarrhea, no systemic toxicity)

QUESTION

| Which of the following are Methanol poisoning derivatives?

A Formic acid + Oxalic acid

B Formic acid + Lactic acid

CORRECT

C Glycolic acid and Oxalic acid

D Oxalic acid and Lactic acid

RIGHT ANSWER

OK B. Formic acid + Lactic acid

WHY THIS IS RIGHT

Methanol poisoning leads to formation of toxic metabolites through alcohol metabolism in the liver:
Methanol → Formaldehyde → Formic acid

Key toxic metabolite:

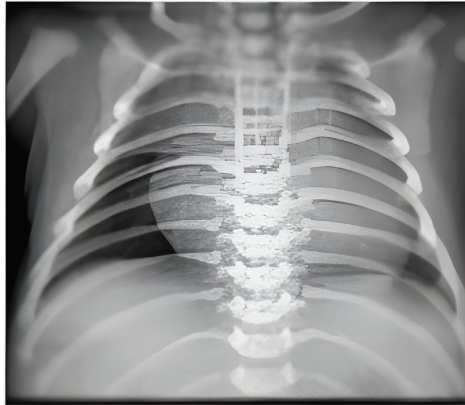
- Formic acid → causes metabolic acidosis and optic nerve toxicity (visual disturbances/blindness)
- Tissue hypoxia due to mitochondrial inhibition → ↑ lactic acid production

Why other options are incorrect:

- Oxalic acid: Seen in ethylene glycol poisoning (causes renal failure, calcium oxalate crystals)
- Glycolic acid: Also from ethylene glycol metabolism

QUESTION

A patient present with an X-ray showing cardiomegaly, along with symptoms of hypotonia, macroglossia, hepatomegaly, and "floppy baby" syndrome. What is the most likely diagnosis?



A Pompe's disease

CORRECT

B Ebstein anomaly

C Transposition of great arteries

D Von Gierke's disease

RIGHT ANSWER

OK **A. Pompe's disease**

WHY THIS IS RIGHT

The combination of cardiomegaly, hypotonia (floppy baby), macroglossia, and hepatomegaly is characteristic of Pompe disease (Glycogen Storage Disease Type II).

Deficiency of acid α -1,4-glucosidase (acid maltase)

- Impaired breakdown of glycogen in lysosomes
- Leads to glycogen accumulation in cardiac and skeletal muscle
- Causes massive cardiomegaly and muscle weakness

Clinical features explained:

- Cardiomegaly $\rightarrow$ heart failure
- Hypotonia $\rightarrow$ "floppy baby"
- Macroglossia
- Hepatomegaly

Why other options are incorrect:

- Ebstein anomaly: Congenital heart defect, not metabolic
- Transposition of great arteries: Cyanotic heart disease
- Von Gierke disease: Severe hypoglycemia + hepatomegaly (no cardiomegaly)

QUESTION

| A patient is being treated with phenobarbitone. Which of the following is inhibited?

A Complex I -> coenzyme Q

CORRECT

B Complex II -> coenzyme Q

C Complex III -> Cyt. C

D Complex IV -> oxygen

RIGHT ANSWER

OK **A. Complex I -> coenzyme Q**

WHY THIS IS RIGHT

Phenobarbitone (barbiturates) can inhibit the electron transport chain at Complex I (NADH dehydrogenase), specifically blocking the transfer of electrons from NADH to coenzyme Q (ubiquinone).

- Inhibits electron flow from NADH -> CoQ
- Prevents proton pumping at Complex I
- Leads to ↓ ATP production via oxidative phosphorylation

Consequences:

- Cells shift toward anaerobic glycolysis
- May result in lactic acidosis in severe toxicity
- CNS depression due to reduced energy availability

Why other options are incorrect:

- Complex II -> CoQ: Inhibited by malonate
- Complex III -> Cytochrome C: Inhibited by antimycin A
- Complex IV -> O₂: Inhibited by cyanide, CO

QUESTION

A 5-year-old girl was washing her doll with shampoo containing rotenone. Her mother noticed her in an unconscious state. Which is inhibited by the above chemical?

A NADH dehydrogenase

CORRECT

B Succinate dehydrogenase

C Cytochrome C

D Cytochrome oxidase

RIGHT ANSWER

OK

A. NADH dehydrogenase

WHY THIS IS RIGHT

Rotenone is a toxin that inhibits Complex I (NADH dehydrogenase) of the electron transport chain (ETC) in mitochondria.

- Blocks transfer of electrons from NADH to coenzyme Q (ubiquinone)
- Prevents proton pumping across inner mitochondrial membrane
- Leads to ↓ ATP production (oxidative phosphorylation failure)

Consequences:

- Cells switch to anaerobic metabolism -> lactic acidosis
- High-energy tissues (brain) are affected first -> unconsciousness

Why other options are incorrect:

- Succinate dehydrogenase (Complex II): Inhibited by malonate
- Cytochrome C: Electron carrier, not directly inhibited here
- Cytochrome oxidase (Complex IV): Inhibited by cyanide and CO

QUESTION

A 6-month-old infant presents with protuberant abdomen, doll-like facies, and recurrent episodes of hypoglycemia. On examination, there is marked hepatomegaly. Laboratory evaluation suggests Von Gierke disease. What is the primary mechanism responsible for the enlarged liver in this condition?

CORRECT

- A** Excess deposition of glycogen within hepatocytes
- B** Accumulation of glucose-6-phosphate due to enzymatic block
- C** Increased levels of glucose-1-phosphate
- D** Defective glycogen primer (glycogenin) formation

RIGHT ANSWER

OK

A. Excess deposition of glycogen within hepatocytes

WHY THIS IS RIGHT

The source quiz contains the placeholder "\$19" here and does not include a written explanation for this item.

QUESTION

| Which of the following activate glycogen phosphorylase?

A Ca²⁺

CORRECT

B Glucose 6-Phosphate

C Glucose

D High ATP

RIGHT ANSWER

OK A. Ca²⁺

WHY THIS IS RIGHT

No explanation was provided in the source quiz.

QUESTION

| Congenital lactic acidosis is due to the defect of

- A** Transketolase
- B** Branched chain ketoacid dehydrogenase
- C** Isocitrate dehydrogenase
- D** **Pyruvate dehydrogenase**

CORRECT

RIGHT ANSWER

- OK** **D. Pyruvate dehydrogenase**

WHY THIS IS RIGHT

Congenital lactic acidosis commonly results from a deficiency of the pyruvate dehydrogenase (PDH) complex, which converts pyruvate to acetyl-CoA, linking glycolysis to the TCA cycle in mitochondria. When PDH is defective, pyruvate cannot enter the TCA cycle and is instead converted to lactate by lactate dehydrogenase, leading to accumulation of lactic acid and metabolic acidosis. Patients often present with neurological deficits, developmental delay, hypotonia, and persistent lactic acidosis. Thus, pyruvate dehydrogenase deficiency impairs aerobic glucose metabolism and causes congenital lactic acidosis.

Why not others:

- Transketolase: Enzyme of the HMP shunt (pentose phosphate pathway), not involved in pyruvate metabolism.
- Branched-chain ketoacid dehydrogenase: Deficiency causes Maple syrup urine disease, not lactic acidosis.
- Isocitrate dehydrogenase: TCA cycle enzyme; defects are mainly associated with certain tumors, not congenital lactic acidosis.

QUESTION

Match the following disorders with their respective clinical features or lab findings.

Disorder	Clinical Feature or Lab Findings
1. Cori's disease	A. Branched structure in liver biopsy
2. McArdle's disease	B. Accumulation of glycogen in lysosome
3. Pompe's disease	C. Exercise intolerance
4. Von Gierke's disease	D. Hypoglycaemia with lactic acidosis

A 1 -> B, 2 -> C, 3 -> A, 4 -> D

B 1 -> C, 2 -> B, 3 -> A, 4 -> D

C 1 -> A, 2 -> C, 3 -> B, 4 -> D

CORRECT

D 1 -> A, 2 -> C, 3 -> D, 4 -> B

RIGHT ANSWER

OK C. 1 -> A, 2 -> C, 3 -> B, 4 -> D

WHY THIS IS RIGHT

No explanation was provided in the source quiz.

Disorder	Finding	Key point
1. Cori's disease	A. Branched structure in liver biopsy	Debranching enzyme deficiency → short outer chains
2. McArdle's disease	C. Exercise intolerance	No rise in lactate with exercise; "second-wind phenomenon"
3. Pompe's disease	B. Glycogen in lysosomes	Only glycogen storage disease that is lysosomal and causes cardiomyopathy
4. Von Gierke's disease	D. Hypoglycaemia + lactic acidosis	G6-phosphatase deficiency → hepatomegaly, doll-face appearance

QUESTION

| Which of the following is not a cytochrome C oxidase inhibitor?

A Carbon monoxide

B Nitric oxide

CORRECT

C Hydrogen sulfide

D Cyanide

RIGHT ANSWER

OK B. Nitric oxide

WHY THIS IS RIGHT

Cytochrome c oxidase (Complex IV) is the final enzyme of the mitochondrial electron transport chain that transfers electrons from cytochrome c to oxygen, forming water and enabling oxidative phosphorylation and ATP production.

Several toxic substances inhibit this enzyme by binding to the heme-iron center, thereby blocking electron transfer to oxygen and halting cellular respiration. Important inhibitors include:

- Cyanide
- Carbon monoxide
- Hydrogen sulfide
- Azide

Inhibition leads to histotoxic hypoxia, where oxygen is present but cells cannot utilize it for ATP generation.

Nitric oxide (NO), although it can interact with mitochondrial enzymes in signaling pathways, is not classically considered a cytochrome c oxidase inhibitor.

QUESTION

A patient presents with hepatosplenomegaly and hypoglycemia. After a meal, glucagon administration increases blood glucose levels. However, after an overnight fast, glucagon administration does not improve blood glucose levels. Liver biopsy reveals increased abnormal glycogen accumulation. Which enzyme deficiency is most likely?

A Muscle phosphorylase

B Glycogen phosphorylase

C Debranching enzyme

CORRECT

D Glucose-6-phosphatase

RIGHT ANSWER

OK C. Debranching enzyme

WHY THIS IS RIGHT

Debranching enzyme deficiency (Glycogen Storage Disease type III - Cori disease) leads to accumulation of abnormally structured glycogen with short outer branches (limit dextrin) in the liver.

Key Clinical Features

- Hepatomegaly and hypoglycemia
- Abnormal glycogen accumulation in liver
- Partial ability to mobilize glycogen

Key Diagnostic Clue

- After a meal: glucagon increases glucose because some glycogen breakdown is still possible.
- After prolonged fasting: glucagon cannot increase glucose, as the remaining glycogen cannot be further degraded due to lack of debranching enzyme.

| Disease | Enzyme Defect | Key Feature |

| ----- | ----- | ----- |

| Von Gierke (Type I) | Glucose-6-phosphatase | Severe fasting hypoglycemia, lactic acidosis |

| Cori disease (Type III) | Debranching enzyme | Limit dextrin accumulation, hepatomegaly |

| McArdle disease (Type V) | Muscle phosphorylase | Exercise intolerance, myoglobinuria |

QUESTION

Carbon monoxide and cyanide cause death primarily due to their interference with which of the following cellular processes?

A Myoglobin

B Cytochrome oxidase

CORRECT

C Hemoglobin

D NADH Dehydrogenase

RIGHT ANSWER

B. Cytochrome oxidase

WHY THIS IS RIGHT

Carbon monoxide and cyanide cause cellular hypoxia by inhibiting the final step of the mitochondrial electron transport chain.

- Cyanide binds to cytochrome c oxidase (Complex IV) in mitochondria.
- This blocks the transfer of electrons to oxygen, preventing formation of water.
- As a result, oxidative phosphorylation stops, ATP production ceases, and cells develop histotoxic hypoxia (oxygen is present but cannot be utilized).

Carbon monoxide (CO) contributes to hypoxia by two mechanisms:

1. High affinity binding to hemoglobin -> formation of carboxyhemoglobin -> reduced oxygen delivery.
- 2\ Binding to mitochondrial cytochrome oxidase -> inhibition of cellular respiration\.

QUESTION

A patient presents with hepatosplenomegaly and a cherry-red spot on the macula. Which enzyme deficiency is most likely responsible for this clinical picture?

A Sphingomyelinase

CORRECT

B Hexosaminidase A

C Glucose 6 phosphatase

D Galactocerebrosidase

RIGHT ANSWER

OK A. Sphingomyelinase

WHY THIS IS RIGHT

Lysosomal Storage Diseases and Associated Enzyme Deficiencies

Disease	Enzyme Deficiency
Tay-Sachs disease	Hexosaminidase A
Sandhoff's disease	Hexosaminidase A and B
Niemann-Pick disease	Sphingomyelinase
Metachromatic leukodystrophy	Arylsulfatase A
Krabbe disease	B-Galactosidase
Gaucher's disease	B-Glucosidase
Fabry's disease	α -Galactosidase
Farber's disease	Ceramidase

QUESTION

| The phenomenon of cancer cells consuming more glucose is due to?

A Lactic acid is produced despite presence of oxygen

CORRECT

B Enhanced conversion of glucose to pyruvate

C Increased pyruvate to acetyl CoA (High NADH/NAD ratio)

D Absence of Oxygen

RIGHT ANSWER

OK

A. Lactic acid is produced despite presence of oxygen

WHY THIS IS RIGHT

Cancer cells preferentially utilize aerobic glycolysis, a phenomenon known as the Warburg effect. Even in the presence of adequate oxygen, cancer cells convert glucose to lactate instead of fully oxidizing pyruvate in mitochondria.

Consequences

- Increased glucose uptake
- Excess lactate production
- Provides biosynthetic intermediates needed for rapid cell proliferation.

QUESTION

A blood sample kept in vitro for metabolic analysis shows reduced glucose utilization along with a paradoxical increase in 2,3-BPG levels within red blood cells. The patient also has features suggestive of a chronic hemolytic state. Which enzyme deficiency best explains these findings?

- A** Hexokinase
- B** Pyruvate kinase
- C** Glucose-6-phosphate dehydrogenase

D Phosphoglycerate kinase

CORRECT

RIGHT ANSWER

OK D. Phosphoglycerate kinase

WHY THIS IS RIGHT

Phosphoglycerate kinase (PGK) deficiency blocks the glycolytic step converting 1,3-bisphosphoglycerate -> 3-phosphoglycerate, reducing ATP generation in red blood cells.

- Accumulation of upstream intermediates
- Increased diversion through the Rapoport-Luebering shunt
- ↑ 2,3-BPG levels
- Reduced glycolytic flux -> decreased glucose utilization in vitro

Elevated 2,3-BPG decreases hemoglobin's oxygen affinity, facilitating oxygen release to tissues but reflecting defective glycolysis in RBCs.

QUESTION

True statements about the HMP shunt:

1. Non-oxidative phase is reversible
2. End products are ribose-5-P, O_2 and NADH
3. No ATP produced in oxidative phase
4. Transketolase requires thiamine

A 1, 3 and 4

CORRECT

B 1 and 2

C 2, 3 and 4

D 1 and 4

RIGHT ANSWER

OK A. 1, 3 and 4

WHY THIS IS RIGHT

The source quiz contains the placeholder "\$1a" here and does not include a written explanation for this item.

QUESTION

A 6-year-old child is brought with progressive abdominal distension. On examination, he has massive hepatomegaly and a doll-like facies with chubby cheeks. There is a history of recurrent episodes of irritability and sweating during fasting, which improve after feeding. The most likely metabolic abnormality in this child is:

- A** Fasting hypoglycemia
- B** Fasting hypoketosis
- C** Decreased hepatic glycogen stores
- D** Exercise-induced muscle cramps

CORRECT

RIGHT ANSWER

- OK** **A. Fasting hypoglycemia**

WHY THIS IS RIGHT

The presentation describes Glycogen Storage Disease Type I (Von Gierke disease) caused by deficiency of glucose-6-phosphatase.

Glucose-6-phosphatase is required to convert glucose-6-phosphate → free glucose in the liver and kidney during:

- Glycogenolysis
- Gluconeogenesis

Typical clinical features:

- Hepatomegaly due to glycogen accumulation
- Doll-like face (fat cheeks)
- Severe fasting hypoglycemia
- Lactic acidosis
- Hyperuricemia
- Hyperlipidemia

Why other options are incorrect:

- Fasting hypoketosis → seen in fatty acid oxidation defects, not Von Gierke disease
- Decreased hepatocellular glycogen → actually glycogen increases in the liver
- Muscle cramps → more typical of McArdle disease (GSD V)

QUESTION

A child is diagnosed with Gaucher's disease. Which statements are true?

1. Deficiency of sphingomyelinase
2. Deficiency of β -glucocerebrosidase
3. Presence of foam cells
4. Crumpled tissue-paper cytoplasm

A 2 and 4

CORRECT

B 1 and 3

C 1, 2 and 4

D 2, 3 and 4

RIGHT ANSWER

OK **A. 2 and 4**

WHY THIS IS RIGHT

Gaucher disease is a lysosomal storage disorder caused by deficiency of the enzyme β -glucocerebrosidase, leading to accumulation of glucocerebroside in macrophages.

- Enzyme deficiency: β -glucocerebrosidase $\rightarrow$ impaired breakdown of glucocerebroside
- Affected cells: macrophages in liver, spleen, bone marrow
- Characteristic histology: macrophages with "crumpled tissue-paper" cytoplasm due to accumulated lipid

These macrophages are called Gaucher cells.

Why other statements are incorrect:

- Deficiency of sphingomyelinase $\rightarrow$ seen in Niemann-Pick disease, not Gaucher disease.
- Foam cells $\rightarrow$ typically described in Niemann-Pick disease due to sphingomyelin accumulation.

QUESTION

Which intermediate of the TCA cycle participates in fatty acid synthesis?

- A** Malate
- B** Fumarate
- C** Succinate
- D** Citrate

CORRECT

RIGHT ANSWER

OK
D. Citrate

WHY THIS IS RIGHT

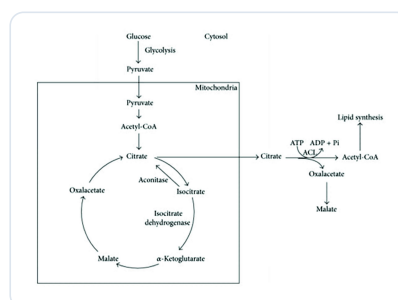
Citrate is the TCA cycle intermediate that links carbohydrate metabolism with fatty acid synthesis.

Role of citrate in fatty acid synthesis

- In the mitochondria, acetyl-CoA enters the TCA cycle and combines with oxaloacetate to form citrate.
- When cellular energy is abundant, citrate is transported from the mitochondria to the cytosol.
- In the cytosol, ATP-citrate lyase cleaves citrate into:
 - Acetyl-CoA
 - Oxaloacetate

The acetyl-CoA produced in the cytosol is the precursor for fatty acid synthesis.

Citrate also Activates acetyl-CoA carboxylase, the rate-limiting enzyme of fatty acid synthesis.



QUESTION

A 6-month-old infant is brought with lethargy, vomiting, and poor feeding after a period of prolonged fasting. Labs show hypoglycemia with low ketone bodies (hypoketosis), suggesting a defect in fat metabolism. Which of the following is the most likely disorder?

- A** Hers disease
- B** Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
- C** Hereditary fructose intolerance
- D** Glycogen storage disease type I (Von Gierke disease)

CORRECT

RIGHT ANSWER

- OK** B. Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency

WHY THIS IS RIGHT

Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency is a fatty acid β -oxidation disorder in which the body cannot properly break down medium-chain fatty acids in mitochondria to generate energy during fasting.

In MCAD deficiency, impaired β -oxidation results in:

- Decreased energy production
- Reduced ketone body formation (hypoketosis)
- Hypoglycemia during fasting

This leads to symptoms such as lethargy, vomiting, poor feeding, and hypoketotic hypoglycemia, especially after prolonged fasting or illness.

Why other options are incorrect:

- Hers disease (GSD VI): Liver glycogen phosphorylase deficiency causing mild hypoglycemia and hepatomegaly, not hypoketotic hypoglycemia.
- Hereditary fructose intolerance: Presents after fructose ingestion with hypoglycemia and vomiting.
- Von Gierke disease (GSD I): Causes severe fasting hypoglycemia with hepatomegaly and lactic acidosis, but ketone production is not characteristically absent.

QUESTION

A patient with family history presents with tendon xanthomas. Laboratory investigations reveal elevated cholesterol with normal triglyceride levels. What is the most likely diagnosis?

- A** Familial hyperlipidemia
- B** Familial dysbetalipoproteinemia
- C** Familial hypertriglyceridemia
- D** **Familial hypercholesterolemia**

CORRECT

RIGHT ANSWER

- OK** **D. Familial hypercholesterolemia**

WHY THIS IS RIGHT

Familial hypercholesterolemia (FH) is an autosomal dominant disorder characterized by defective or absent LDL receptors, leading to impaired clearance of low-density lipoprotein (LDL) cholesterol from the blood. This results in marked elevation of LDL cholesterol with normal triglyceride levels.

The excess cholesterol gets deposited in tissues, producing characteristic findings such as:

- Tendon xanthomas (commonly in Achilles tendon and extensor tendons)
- Xanthelasma
- Premature atherosclerosis and early coronary artery disease

Why other options are incorrect:

- Familial hyperlipidemia: A nonspecific term and not a defined disorder.
- Familial dysbetalipoproteinemia: Characterized by elevation of both cholesterol and triglycerides with palmar xanthomas.
- Familial hypertriglyceridemia: Mainly shows elevated triglycerides, not isolated high cholesterol.

QUESTION

Fatty acid transport through mitochondrial membrane is via?

CORRECT

A

Carnitine

B

Acyl carrier protein

C

Cholesterol transporter

D

LCAT

RIGHT ANSWER

OK

A. Carnitine

WHY THIS IS RIGHT

Long-chain fatty acids cannot directly cross the inner mitochondrial membrane. Their transport into the mitochondrial matrix for β -oxidation requires the carnitine shuttle system.

Mechanism (Carnitine shuttle):

1. Fatty acids are activated to fatty acyl-CoA in the cytosol.
2. Carnitine palmitoyltransferase I (CPT-I) converts acyl-CoA to acyl-carnitine (outer mitochondrial membrane).
3. Acyl-carnitine is transported across the inner membrane via a translocase.
4. CPT-II reconverts acyl-carnitine to acyl-CoA inside the mitochondria for β -oxidation.

Defects in the carnitine shuttle (e.g., CPT deficiency) lead to impaired fatty acid oxidation $\rightarrow$ hypoketotic hypoglycemia and muscle weakness.

Why other options are incorrect:

- Acyl carrier protein: Involved in fatty acid synthesis, not transport.
- Cholesterol transporter: Not related to fatty acid mitochondrial entry.
- LCAT (Lecithin-cholesterol acyltransferase): Involved in cholesterol metabolism (HDL function).

QUESTION

A patient has multiple tendon xanthomas. Serum cholesterol (398 mg/dL) and LDL 220 mg/dL were found to be raised. What is the defect?

A Lipoprotein lipase deficiency

B LDL receptors defect

CORRECT

C Apo E defect

D LCAT deficiency

RIGHT ANSWER

B. LDL receptors defect

WHY THIS IS RIGHT

The presence of tendon xanthomas with markedly elevated total cholesterol and LDL is characteristic of familial hypercholesterolemia (Type IIa).

- Mutation in LDL receptor -> impaired uptake of LDL by liver
- Leads to accumulation of LDL in plasma
- Normal triglycerides

Pathophysiology:

- Excess LDL deposits cholesterol in tissues -> tendon xanthomas
- Promotes atherosclerosis -> premature coronary artery disease

Why other options are incorrect:

- Lipoprotein lipase deficiency: ↑ triglycerides (chylomicrons), not LDL
- Apo E defect: Dysbetalipoproteinemia (↑ cholesterol + TG, palmar xanthomas)
- LCAT deficiency: HDL metabolism defect (fish-eye disease), not tendon xanthomas

QUESTION

A patient came to the emergency room with severe pain in the abdomen. Serum triglyceride level was 1500 mg/dL. Identify the defect?

- A Apo B 48
- B Apo B 100
- C Apo CII**
- D LDL receptor

CORRECT

RIGHT ANSWER

- C. Apo CII**

WHY THIS IS RIGHT

Very high triglycerides (>1000 mg/dL) with acute abdominal pain suggests hypertriglyceridemia-induced pancreatitis, classically seen in familial chylomicronemia (Type I hyperlipoproteinemia).

- Deficiency of Apolipoprotein C-II (Apo C-II)
- Apo C-II is a cofactor for lipoprotein lipase (LPL)

Pathophysiology:

- Without Apo C-II -> LPL cannot be activated
- Impaired breakdown of chylomicrons -> massive accumulation of triglycerides
- Leads to pancreatitis, eruptive xanthomas, lipemia retinalis

Why other options are incorrect:

- Apo B-48: Needed for chylomicron formation (defect -> abetalipoproteinemia)
- Apo B-100: Required for LDL/VLDL metabolism (familial hypercholesterolemia-like states)
- LDL receptor: Causes ↑ LDL (Type IIa), not severe triglyceridemia

QUESTION

A patient presents with xanthomas on the Achilles tendon. Which of the following is the diagnosis?

- A** Tangier's disease
- B** Familial hyperchylomicronemia
- C** **Familial hypercholesterolemia**
- D** Familial Dysbetalipoproteinemia

CORRECT

RIGHT ANSWER

- OK** **C. Familial hypercholesterolemia**

WHY THIS IS RIGHT

Tendon xanthomas (classically over the Achilles tendon) are a hallmark of familial hypercholesterolemia (Type II hyperlipoproteinemia). This condition is caused by a defect in LDL receptors, leading to markedly elevated LDL cholesterol levels and deposition of cholesterol in tendons and skin. It is associated with premature atherosclerosis and early coronary artery disease.

Key differentiation:

- Familial hypercholesterolemia: Tendon xanthomas
- Familial dysbetalipoproteinemia: Palmar xanthomas
- Hyperchylomicronemia: Eruptive xanthomas
- Tangier disease: Low HDL, orange tonsils

QUESTION

Patient with high triglycerides esterified with long chain fatty acid (LCFA) presents with fatigue and biopsy of muscle shows fat vacuoles. Which of the following is the diagnosis?

A Carnitine deficiency

CORRECT

B FA synthase defect

C LPL defect

D LDL receptor defect

RIGHT ANSWER

OK

A. Carnitine deficiency

WHY THIS IS RIGHT

Carnitine deficiency is a disorder of fatty acid oxidation in which long-chain fatty acids (LCFA) cannot be transported into mitochondria for β -oxidation. Carnitine is required for the carnitine shuttle

- LCFA cannot be utilized -> accumulate as triglycerides in muscle
- Leads to lipid (fat) vacuoles in muscle biopsy
- $\downarrow$ energy production -> especially during fasting/exercise

Clinical features:

- Muscle weakness & fatigue
- Hypoketotic hypoglycemia
- Myopathy with lipid accumulation

Why other options are incorrect:

- Fatty acid synthase defect: Affects synthesis, not oxidation
- Lipoprotein lipase (LPL) defect: Causes $\uparrow$ triglycerides in plasma (chylomicronemia), not muscle fat vacuoles
- LDL receptor defect: Causes hypercholesterolemia, not fatty acid oxidation defect

QUESTION

A patient presents with seizures, a cherry-red spot on the macula, and no hepatosplenomegaly. Laboratory investigations reveal a deficiency of hexosaminidase. Which compound is accumulated in this patient?

A Galactosylceramide

B GM2 ganglioside

CORRECT

C Sphingomyelin

D GM1 ganglioside

RIGHT ANSWER

OK B. GM2 ganglioside

WHY THIS IS RIGHT

The clinical triad of seizures, neurodegeneration, cherry-red spot on macula, and absence of hepatosplenomegaly is characteristic of Tay-Sachs disease.

- Deficiency of hexosaminidase A
- Leads to accumulation of GM2 ganglioside in neurons

Pathophysiology:

- GM2 accumulates in lysosomes of neurons -> progressive neurodegeneration
- Retina becomes pale due to lipid accumulation, while the fovea appears as a cherry-red spot
- No organomegaly (important differentiating feature)

Why other options are incorrect:

- Galactosylceramide: Accumulates in Krabbe disease
- Sphingomyelin: Accumulates in Niemann-Pick disease (with hepatosplenomegaly)
- GM1 ganglioside: Accumulates in GM1 gangliosidosis

QUESTION

A 1-year-old child is brought with developmental delay, progressive abdominal distension, and recurrent respiratory infections. On examination, the child has coarse facial features, hepatosplenomegaly, corneal clouding, and skeletal abnormalities. Enzyme assay reveals deficiency of α -L-iduronidase, consistent with Hurler syndrome. Which of the following substances accumulates in this condition?

A Heparan sulfate (HS) and Dermatan sulfate (DS)

CORRECT

B Chondroitin sulfate (CS) and Keratan sulfate (KS)

C Dermatan sulfate (DS) only

D Sphingolipids

RIGHT ANSWER

OK A. Heparan sulfate (HS) and Dermatan sulfate (DS)

WHY THIS IS RIGHT

The clinical picture-coarse facial features, organomegaly, corneal clouding, and intellectual disability-with deficiency of α -L-iduronidase is characteristic of Hurler syndrome (Mucopolysaccharidosis type I).

- α -L-iduronidase is required for degradation of glycosaminoglycans (GAGs)
- Deficiency leads to accumulation of:
 - o Heparan sulfate (HS)
 - o Dermatan sulfate (DS)

Mucopolysaccharidoses (MPS):

Enzyme Defect	Mucopolysaccharidosis	Enzyme Deficiency	GAGs Accumulated	Key Symptoms
Hurler Syndrome Hurler-Scheie Syndrome	Hurler-Scheie Syndrome	α -L-Iduronidase	Dermatan Sulfate, Heparan Sulfate	Coarse facial features, corneal clouding, hepatosplenomegaly, skeletal abnormalities
Hunter Syndrome	Hunter Syndrome	Iduronate sulfatase	Dermatan Sulfate, Heparan Sulfate	Mental retardation
Sandhoff Syndrome A	Sandhoff Syndrome A	Heparan sulfate 6-O-sulfatase (Heparanase)	Heparan Sulfate	Developmental delay, mental retardation
Sandhoff Syndrome B	Sandhoff Syndrome B	α -N-Acetylglucosaminidase	Heparan Sulfate	Similar to MPS IIA
Sandhoff Syndrome C	Sandhoff Syndrome C	α -Glucuronidase	Heparan Sulfate	Similar to MPS IIA
Sandhoff Syndrome D	Sandhoff Syndrome D	N-Acetylglucosaminase 6-sulfatase	Heparan Sulfate	Similar to MPS IIA
Morquio Syndrome A	Morquio Syndrome A	Glucosaminase 6-sulfatase	Keratan Sulfate, Chondroitin 6-Sulfate	Mental retardation, short stature
Morquio Syndrome B	Morquio Syndrome B	B-Galactosidase	Keratan Sulfate	Similar to MPS IIA
Hurler-Scheie-Langer Syndrome	Hurler-Scheie-Langer Syndrome	N-Acetylglucosaminase 6-sulfatase (Heparanase II)	Dermatan Sulfate, Heparan Sulfate, Chondroitin 6-Sulfate	Mental retardation, short stature, skeletal abnormalities
By Syndrome	By Syndrome	α -Chondroitinase	Dermatan Sulfate, Heparan Sulfate, Chondroitin 6-Sulfate	Mental retardation, short stature, skeletal abnormalities
Hurler-Scheie Syndrome	Hurler-Scheie Syndrome	Hydrolase	Hydroxyacid	Joint pain, short stature

QUESTION

A patient presents with xanthomas and elevated cholesterol levels. Laboratory investigations show high LDL but normal triglyceride (TG) levels. There is also a family history of premature myocardial infarction (MI)-the patient's father died due to MI. What is the most likely diagnosis?

A Hyperlipoproteinemia III

B Hyperlipoproteinemia I

C Hyperlipoproteinemia IIa

CORRECT

D Hyperlipoproteinemia IIb

RIGHT ANSWER

OK C. Hyperlipoproteinemia IIa

WHY THIS IS RIGHT

The clinical picture-tendon xanthomas, elevated LDL cholesterol with normal triglycerides, and family history of premature myocardial infarction-is characteristic of Type IIa hyperlipoproteinemia (familial hypercholesterolemia).

- Mutation in LDL receptor (most common) or ApoB-100
- Leads to reduced clearance of LDL from plasma

Lipid profile:

- ↑ LDL cholesterol
- Normal triglycerides

Clinical features:

- Tendon xanthomas (Achilles, extensor tendons)
- Xanthelasma
- Premature atherosclerosis -> early MI

Why other options are incorrect:

- Type III (dysbetalipoproteinemia): ↑ cholesterol + ↑ triglycerides, palmar xanthomas
- Type I: ↑ chylomicrons -> ↑ triglycerides (pancreatitis)
- Type IIb: ↑ LDL + ↑ VLDL -> ↑ cholesterol + ↑ triglycerides

QUESTION

A patient presented with orange coloured tonsils, decreased HDL <5 mg/dl, triglyceride 140 mg/dl. What is the most likely diagnosis?

A Tangier disease

CORRECT

B Abetalipoproteinemia

C Familial hypercholesterolemia

D Familial hyperchylomicronemia

RIGHT ANSWER

OK A. Tangier disease

WHY THIS IS RIGHT

Tangier disease is a rare autosomal recessive disorder caused by a defect in the ABCA1 transporter, which is responsible for efflux of cholesterol from peripheral cells to HDL.

- Impaired cholesterol transport -> failure of HDL formation
- Leads to extremely low HDL levels (<5 mg/dL)
- Cholesterol accumulates in tissues of the reticuloendothelial system

Characteristic clinical features:

- Orange-colored tonsils (pathognomonic clue)
- Very low HDL levels
- Peripheral neuropathy
- Hepatosplenomegaly (sometimes)

Why other options are incorrect:

- Abetalipoproteinemia: Absence of Apo B -> fat malabsorption, acanthocytosis
- Familial hypercholesterolemia: ↑ LDL, tendon xanthomas
- Familial hyperchylomicronemia: ↑ triglycerides, pancreatitis

QUESTION

Which of the following defects causes familial hypercholesterolemia?

A Apo E

B Apo CII

C LDL receptor

CORRECT

D Apo B48

RIGHT ANSWER

OK

C. LDL receptor

WHY THIS IS RIGHT

Familial hypercholesterolemia (FH) is most commonly caused by a defect in the LDL receptor, which is responsible for removing low-density lipoprotein (LDL) particles from the bloodstream. When the LDL receptor is defective or absent, LDL cannot be efficiently taken up by the liver, leading to markedly elevated plasma LDL cholesterol levels. This results in premature atherosclerosis, tendon xanthomas, and an increased risk of early coronary artery disease.

Other options are associated with different lipid disorders:

- Apo E defect -> Type III hyperlipoproteinemia
- Apo CII defect -> Impaired lipoprotein lipase activation (Type I hyperlipoproteinemia)
- Apo B48 -> Involved in chylomicron formation, not FH.

Type	Inherit	Pathogenesis	Blood Level	Clinical
I- Hyper- -chylomicronaemia	Ar	Lipoprotein Lipase/ApoC-II deficiency	Chylomicrons, TG, Cholesterol	• Pancreatitis • Eruptive xanthomas • CREAM Supernatant
II- Familial hyper cholesterolemia	AD	Absent LDL Receptors/ ApoB-100	IIa: LDL IIb: LDL + TG	• Accelerated Atherosclerosis • Tendon Xanthomas • Corneal arcus, Xanthosoma (Hb) • Tubero-eruptive xanthomas (Hb)
III- Dysbeta- lipoproteinemia	Ar	Defective ApoE	Chylomicrons, VLDL, LDL	• Premature atherosclerosis • Tubero-Eruptive & Palmar Xanthomas • Broad beta band
IV- Hyper- triglyceridemia	AD	↑↑↑ Hepatic VLDL	VLDL, TG Type 5: VLDL + chylomicrons	• Broad beta band • Acute pancreatitis • Insulin resistance • Tubero-Eruptive Xanthomas

QUESTION

Statement NOT true about β -oxidation of fatty acids:

- A** CAT I and CAT II transport fatty acids
- B** ATP is used for transport
- C** CoASH released in cytosol during acyl-carnitine formation CORRECT
- D** Carnitine released inside mitochondria

RIGHT ANSWER

- OK** **C. CoASH released in cytosol during acyl-carnitine formation**

WHY THIS IS RIGHT

β -oxidation of fatty acids occurs in the mitochondrial matrix, and long-chain fatty acids require the carnitine shuttle for mitochondrial transport.

Carnitine Shuttle Steps:

1. CPT-I (CAT-I): Converts fatty acyl-CoA $\rightarrow$ acyl-carnitine in the cytosol/outer mitochondrial membrane.
2. Acyl-carnitine is transported into mitochondria.
3. CPT-II (CAT-II): Converts acyl-carnitine $\rightarrow$ fatty acyl-CoA in the mitochondrial matrix, releasing carnitine.

Key points:

- ATP is used earlier for fatty acid activation to fatty acyl-CoA.
- Carnitine is released inside mitochondria after reconversion.
- CoA remains within the mitochondrial matrix, not released in the cytosol during acyl-carnitine formation.

QUESTION

A 17-year-old tall male presents for a routine check-up. He has long limbs, arachnodactyly, hypermobile joints, a high-arched palate, and an increased arm span. Genetic testing reveals a defect in the FBN1 gene. Which of the following is the most likely diagnosis?

- A** Ehlers-Danlos Syndrome
- B** Marfan Syndrome
- C** Osteogenesis Imperfecta
- D** Heritable Disorders of Connective Tissue

CORRECT

RIGHT ANSWER

B. Marfan Syndrome

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$19” here and does not include a written explanation for this item.

QUESTION

| Respiratory distress syndrome is caused by the deficiency of?

- A** Cephalin
- B** Sphingomyelin
- C** **Lecithin**
- D** Cardiolipin

CORRECT

RIGHT ANSWER

OK **C. Lecithin**

WHY THIS IS RIGHT

Respiratory Distress Syndrome (RDS) in newborns is caused by deficiency of pulmonary surfactant, the most important component of which is lecithin (dipalmitoyl phosphatidylcholine, DPPC).

Function of lecithin (surfactant):

- Reduces alveolar surface tension
- Prevents alveolar collapse (atelectasis) during expiration
- Maintains lung compliance

Clinical correlation:

- Common in premature infants (immature type II pneumocytes)
- Assessed by Lecithin : Sphingomyelin (L/S) ratio in amniotic fluid

o L/S ratio < 2 -> high risk of RDS

Why other options are incorrect:

- Cephalin: Phospholipid involved in coagulation
- Sphingomyelin: Used as a reference in L/S ratio, not the main surfactant
- Cardiolipin: Found in mitochondrial membranes

QUESTION

An infant is brought by his parents with complaints that his urine turns black on standing. Which of the following metabolic disorders is likely?

- A Phenylketonuria
- B Alkaptonuria**
- C Homocystinuria
- D Maple syrup urine disease

CORRECT

RIGHT ANSWER

- B. Alkaptonuria**

WHY THIS IS RIGHT

Alkaptonuria is an autosomal recessive disorder of tyrosine metabolism caused by deficiency of the enzyme homogentisate oxidase.

Clinical features:

- Dark/black urine on standing (early clue in infants)
- Ochronosis (bluish-black pigmentation of connective tissue)
- Arthritis in adulthood

Why other options are incorrect:

- Phenylketonuria (PKU): Musty/mousy odor urine, intellectual disability
- Homocystinuria: Marfanoid habitus, lens dislocation (downward)
- Maple syrup urine disease: Sweet “maple syrup” odor of urine

QUESTION

An adult patient presents with tall stature, pigeon chest deformity, long arm span, lens dislocation, and cardiac defects. This patient is likely to have a defect in which of the following?

A Elastase

B Fibrillin

CORRECT

C Collagen

D Laminin

RIGHT ANSWER

OK

B. Fibrillin

WHY THIS IS RIGHT

The clinical features-tall stature, long arm span, pectus deformity (pigeon chest), lens dislocation, and cardiac abnormalities (aortic root dilation)-are classic for Marfan syndrome.

- Mutation in fibrillin-1 (FBN1 gene)
- Fibrillin is a key component of microfibrils in connective tissue

Clinical features explained:

- Skeletal: Tall, long limbs (arachnodactyly), pectus deformity
- Ocular: Lens dislocation (typically upward)
- Cardiac: Aortic root dilation -> risk of aortic aneurysm/dissection

Why other options are incorrect:

- Elastase: Enzyme, not structural protein defect
- Collagen: Seen in Ehlers-Danlos or osteogenesis imperfecta
- Laminin: Basement membrane component

QUESTION

A 26-year-old male presented with colicky pain. Following type of crystals were found on urine examination. What is the likely diagnosis?



- A** ADPKD
- B** Calcium oxalate stones
- C** Cystinuria
- D** Glomerulonephritis

CORRECT

RIGHT ANSWER

OK C. Cystinuria

WHY THIS IS RIGHT

The presence of hexagonal crystals in urine is pathognomonic of cystinuria, a genetic disorder affecting amino acid transport. Defective renal tubular reabsorption of cystine and dibasic amino acids (COLA: cystine, ornithine, lysine, arginine)

- Cystine is poorly soluble in urine -> precipitates to form hexagonal crystals and stones
- Leads to recurrent renal colic and kidney stones

Clinical features:

- Recurrent kidney stones at a young age
- Colicky abdominal pain
- May have family history (autosomal recessive)

Why other options are incorrect:

- Calcium oxalate stones: Envelope-shaped crystals
- ADPKD: Cystic kidneys, not crystal-related
- Glomerulonephritis: Hematuria, proteinuria (no characteristic crystals)

QUESTION

Which of the following is deficient in a patient with phenylketonuria with normal phenylalanine hydroxylase levels?

A Dihydrobiopterin reductase

CORRECT

B Tyrosine transaminase

C Dopa decarboxylase

D Tyrosine hydroxylase

RIGHT ANSWER

OK A. Dihydrobiopterin reductase

WHY THIS IS RIGHT

In a patient with hyperphenylalaninemia but normal phenylalanine hydroxylase (PAH), the defect lies in the cofactor tetrahydrobiopterin (BH₄) metabolism.

Key enzyme involved: Dihydrobiopterin reductase (DHPR) Deficiency -> ↓ BH₄ availability

- Impaired phenylalanine -> tyrosine conversion -> ↑ phenylalanine
- Also affects neurotransmitter synthesis:

- o ↓ dopamine (tyrosine pathway)
- o ↓ serotonin (tryptophan pathway)

Clinical importance:

- More severe than classical PKU
- Presents with neurological deficits due to neurotransmitter deficiency

Why other options are incorrect:

- Tyrosine transaminase: Involved in tyrosine metabolism (tyrosinemia)
- Dopa decarboxylase: Involved in dopamine synthesis, not primary cause here
- Tyrosine hydroxylase: Requires BH₄ but is not the defective enzyme in this condition

QUESTION

A patient has markedly elevated plasma phenylalanine levels and low tyrosine levels. Phenylalanine hydroxylase enzyme assay is normal. Which of the following cofactor deficiencies is most likely responsible for this presentation?

A Bioppterin

CORRECT

B DHFR

C PLP

D THF

RIGHT ANSWER

OK A. Bioppterin

WHY THIS IS RIGHT

Conversion of phenylalanine → tyrosine requires:

- Enzyme: Phenylalanine hydroxylase (PAH)
- Cofactor: Tetrahydrobiopterin (BH₄, bioppterin)

In this case, PAH is normal, so the defect lies in the cofactor (BH₄) deficiency, leading to:

- ↑ Phenylalanine
- ↓ Tyrosine

BH₄ is also required for synthesis of neurotransmitters:

- Dopamine (via tyrosine hydroxylase)
- Serotonin (via tryptophan hydroxylase)

Thus, deficiency leads to: Neurological symptoms more severe than classical PKU

Why other options are incorrect:

- DHFR: Involved in folate metabolism
- PLP (Vitamin B6): Cofactor for transamination reactions
- THF: Involved in one-carbon metabolism (DNA synthesis)

QUESTION

2, 3 BPG binds to:

A Haem part of Hb

B Globin part of Hb

CORRECT

C Haem part of Myoglobin

D Globin part of Myoglobin

RIGHT ANSWER

OK **B. Globin part of Hb**

WHY THIS IS RIGHT

2,3-Bisphosphoglycerate (2,3-BPG) binds to the positively charged pocket formed by the β -globin chains of deoxyhemoglobin, not to the heme group. This interaction stabilizes the T (tense) state of hemoglobin, thereby reducing hemoglobin's affinity for oxygen and promoting oxygen release to peripheral tissues.

Why other options are incorrect:

- Heme part of Hb: Oxygen binds to the iron in the heme group; 2,3-BPG does not bind here.
- Heme part of Myoglobin: Myoglobin has a single heme and does not interact with 2,3-BPG.
- Globin part of Myoglobin: Myoglobin lacks the central cavity present in hemoglobin and therefore cannot bind 2,3-BPG.

QUESTION

| Which diagnostic tool is preferred for metabolic disease screening in children?

A Tandem mass spectrometry

CORRECT

B Complete blood count (CBC)

C Urinalysis

D Genetic testing

RIGHT ANSWER

OK

A. Tandem mass spectrometry

WHY THIS IS RIGHT

Tandem mass spectrometry (MS/MS) is the preferred screening tool for inborn errors of metabolism in newborns and children. It can simultaneously detect multiple metabolic disorders by measuring amino acids, organic acids, and acylcarnitines from a single dried blood spot.

This method is widely used in newborn screening programs to identify disorders such as phenylketonuria, maple syrup urine disease, fatty acid oxidation defects, and organic acidemias, allowing early diagnosis and treatment to prevent severe complications.

Why not other options:

- CBC: Evaluates blood cells (RBCs, WBCs, platelets); not useful for detecting metabolic enzyme defects.
- Urinalysis: May detect some metabolites but is not sensitive or comprehensive for screening multiple metabolic disorders.
- Genetic testing: Confirms specific mutations but is not practical for large-scale initial screening of many metabolic diseases.

QUESTION

| Which of the following is not an amino acid-derived neurotransmitter?

A Dopamine

B Serotonin

C GABA

D **Creatine**

CORRECT

RIGHT ANSWER

OK **D. Creatine**

WHY THIS IS RIGHT

Many neurotransmitters are derived from amino acids or their metabolites.

- Dopamine is synthesized from the amino acid tyrosine.
- Serotonin (5-HT) is derived from tryptophan.
- GABA (γ -aminobutyric acid) is produced from glutamate by the enzyme glutamate decarboxylase.

Creatine, however, is not a neurotransmitter. It is a high-energy storage molecule synthesized from glycine, arginine, and methionine and is mainly involved in energy metabolism in muscle and brain.

QUESTION

Which is not a product of heme catabolism?

A Ferrous ion

B Aminolaevulinic acid

CORRECT

C Biliverdin

D Carbon monoxide

RIGHT ANSWER

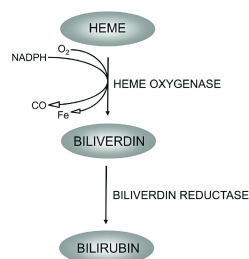
OK B. Aminolaevulinic acid

WHY THIS IS RIGHT

During heme catabolism, heme is degraded by the enzyme heme oxygenase to produce biliverdin, ferrous iron (Fe^{2+}), and carbon monoxide (CO). Biliverdin is subsequently reduced to bilirubin by biliverdin reductase.

Aminolevulinic acid (ALA) is not a product of heme breakdown; instead, it is an intermediate in heme synthesis, formed from glycine and succinyl-CoA by ALA synthase in mitochondria.

Therefore, aminolevulinic acid belongs to the heme biosynthesis pathway, not the catabolic pathway, making it the correct answer.



QUESTION

| Match the following Amino acid products:

A. Tyrosine	1. Melatonin
B. Tryptophan	2. Glutathione
C. Glycine	3. Nor epinephrine
D. Histidine	4. Histamine

A A-2, B-4, C-3, D-1

B A-3, B-1, C -2, D-4

CORRECT

C A-1, B-2, C-3, D-4

D A-4, B-3, C-2, D-1

RIGHT ANSWER

OK B. A-3, B-1, C -2, D-4

WHY THIS IS RIGHT

Amino Acid Conversions

Tyrosine (A)	Norepinephrine (3)
Tryptophan (B)	Melatonin (1)
Glycine (C)	Glutathione (2)
Histidine (D)	Histamine (4)

QUESTION

Sodium benzoate is given for urea cycle defects for scavenging ammonia. Which amino acid donates amino group to the scavengers?

A Arginine

B Glycine

CORRECT

C Glutamine

D Aspartate

RIGHT ANSWER

OK

B. Glycine

WHY THIS IS RIGHT

In urea cycle disorders, toxic ammonia accumulates because the body cannot efficiently convert ammonia to urea. Nitrogen scavenger drugs such as sodium benzoate are used to remove excess nitrogen through alternative metabolic pathways.

Mechanism of Sodium Benzoate:

- Sodium benzoate conjugates with glycine in the liver.
- This reaction forms hippuric acid (benzoylglycine).
- Hippuric acid is then excreted in urine, removing one nitrogen atom from the body.

Drug	Amino acid used	Product excreted
---	-----	-----
Sodium benzoate	Glycine	Hippurate
Sodium phenylacetate / phenylbutyrate	Glutamine	Phenylacetylglutamine

QUESTION

Match the following amino acids with their respective derivatives:

1. Tyrosine
2. Tryptophan
3. Glycine
4. Cysteine
- a. Thyroxine
- b. Taurine
- c. Creatine
- d. Melatonin

A 1-a, 2-d, 3-c, 4-b

CORRECT

B 1-d, 2-a, 3-b, 4-c

C 1-c, 2-b, 3-d, 4-a

D 1-b, 2-c, 3-a, 4-d

RIGHT ANSWER

OK A. 1-a, 2-d, 3-c, 4-b

WHY THIS IS RIGHT

Amino Acid	Important Derivative	Key Function
Tyrosine	Thyroxine (T4)	Thyroid hormone involved in regulation of metabolism
Tryptophan	Melatonin	Hormone regulating circadian rhythm and sleep
Glycine	Creatine	Involved in energy storage in muscles (phosphocreatine system)
Cysteine	Taurine	Important component of bile salts and antioxidant functions

- Tyrosine -> Thyroid hormones (T3, T4), catecholamines, melanin
- Tryptophan -> Serotonin -> Melatonin
- Glycine + Arginine + Methionine -> Creatine
- Cysteine -> Taurine -> Bile salts

QUESTION

A 15-year-old boy presents with tremors, difficulty in speech, and behavioral changes. On examination, he has hepatomegaly and a golden-brown ring at the limbus of the cornea (Kayser-Fleischer ring). Wilson's disease is suspected. Which of the following is the best investigation to support the diagnosis?

- A** Serum ceruloplasmin
- B** Liver biopsy for copper estimation
- C** Serum copper
- D** Copper excretion in urine

CORRECT

RIGHT ANSWER

- OK** **B. Liver biopsy for copper estimation**

WHY THIS IS RIGHT

Wilson disease is an autosomal recessive disorder of copper metabolism caused by defective copper transport and impaired biliary excretion. This results in excess copper accumulation in the liver, brain (basal ganglia), and cornea, producing features such as hepatomegaly, neurological symptoms, behavioral changes, and Kayser-Fleischer rings.

The most definitive investigation to support the diagnosis is liver biopsy for quantitative copper estimation, which directly measures hepatic copper concentration. Markedly increased hepatic copper confirms abnormal copper accumulation.

Why other options are incorrect:

- Serum ceruloplasmin: Often decreased in Wilson disease but can be normal in some cases, so it is not definitive.
- Serum copper: Usually low due to reduced ceruloplasmin-bound copper and is not reliable for diagnosis.
- Urinary copper excretion: Increased in Wilson disease and useful for screening/monitoring but less definitive than hepatic copper estimation.

QUESTION

A 48-year-old male who has been working in a paint manufacturing factory for over 20 years presents with chronic abdominal pain, constipation, fatigue, headache, and joint pains. He also reports irritability and sleep disturbances. On examination, pallor is noted. Laboratory investigations reveal microcytic anemia. Peripheral smear shows basophilic stippling of RBCs. Which of the following enzymes is most likely inhibited in this condition?

- A** Uroporphyrinogen III synthase
- B** Porphobilinogen synthase (ALA dehydratase)
- C** ALA synthase
- D** **Ferrochelatase**

CORRECT

RIGHT ANSWER

- OK** **D. Ferrochelatase**

WHY THIS IS RIGHT

Chronic exposure to lead (e.g., in paint factory workers) causes lead poisoning, which interferes with heme synthesis.

Lead inhibits two key enzymes in the heme pathway:

- Ferrochelatase -> blocks incorporation of iron into protoporphyrin IX to form heme
- ALA dehydratase (ALAD) -> inhibits conversion of ALA to porphobilinogen

Clinical features (classic):

- Abdominal pain (lead colic)
- Constipation
- Neuropathy (wrist/foot drop)
- Anemia
- Irritability, insomnia, headache

Why other options are incorrect:

- Uroporphyrinogen III synthase: Defect causes congenital erythropoietic porphyria
- Uroporphyrinogen I synthase (incorrect term): Not a standard enzyme
- ALA synthase: Rate-limiting enzyme; not inhibited by lead (actually regulated by heme)

QUESTION

A child presents with erythematous scaly patches in the perioral region, mucosal ulcers, and impaired epithelial wound healing. What is the likely mineral deficiency associated with this condition?

A Iron deficiency

B Zinc deficiency

CORRECT

C Calcium deficiency

D Copper deficiency

RIGHT ANSWER

OK B. Zinc deficiency

WHY THIS IS RIGHT

Zinc deficiency leads to characteristic periorificial dermatitis (around mouth, eyes, anus), mucosal ulcers, and impaired wound healing, as seen in this child. Zinc is a cofactor for numerous enzymes involved in:

- DNA & RNA synthesis
- Cell division and tissue repair
- Protein synthesis and epithelial integrity

Deficiency results in: Acrodermatitis enteropathica (genetic zinc malabsorption)

- Impaired epithelial regeneration -> erythematous scaly lesions
- Poor wound healing
- Mucosal breakdown -> ulcers

Other features of zinc deficiency:

- Alopecia
- Diarrhea
- Growth retardation
- Loss of taste (hypogeusia)

Why other options are incorrect:

- Iron deficiency: Causes anemia, not characteristic dermatitis
- Calcium deficiency: Affects bones (rickets/osteomalacia)
- Copper deficiency: Causes anemia and neurologic issues, not perioral dermatitis

QUESTION

A 2-year-old child is brought with progressive pallor, recurrent infections, and failure to thrive. On examination, the child has hypopigmented skin and sparse, light-colored hair. Laboratory investigations reveal anemia and neutropenia. Which of the following micronutrient deficiencies best explains this clinical presentation?

A Zinc

B Selenium

C Iron

D Copper

CORRECT

RIGHT ANSWER

OK

D. Copper

WHY THIS IS RIGHT

The combination of anemia + neutropenia + hypopigmentation of skin and hair is characteristic of copper deficiency.

Copper is a cofactor for several important enzymes:

- Ceruloplasmin -> involved in iron metabolism ($\text{Fe}^{2+} \rightarrow \text{Fe}^{3+}$)
- Cytochrome c oxidase -> energy production
- Tyrosinase -> melanin synthesis

Pathophysiology:

- ↓ Ceruloplasmin -> impaired iron mobilization -> anemia
- Impaired immune function -> neutropenia
- ↓ Tyrosinase activity -> ↓ melanin -> hypopigmentation

Clinical associations:

- Malnutrition
- Excess zinc intake (interferes with copper absorption)
- Menkes disease (genetic copper deficiency)

Why other options are incorrect:

- Zinc: Causes dermatitis, delayed wound healing
- Selenium: Cardiomyopathy (Keshan disease)
- Iron: Causes anemia only (no neutropenia or hypopigmentation)

QUESTION

A patient's relatives sent a message on social media to the consulting doctor, unaware about the other investigations. But he mentioned that urinary coproporphyrin is positive. What is the probable cause?

A Lead poisoning

CORRECT

B Asbestosis

C Silicosis

D Mercury poisoning

RIGHT ANSWER

OK

A. Lead poisoning

WHY THIS IS RIGHT

Lead poisoning interferes with heme synthesis, leading to accumulation of porphyrin precursors such as coproporphyrin in urine. Lead inhibits key enzymes in the heme pathway:

- ALA dehydratase (ALAD)
- Ferrochelatase

Consequences:

- ↑ ALA and coproporphyrin in urine
- ↓ heme synthesis → anemia

Clinical features:

- Abdominal pain (lead colic)
- Neurological symptoms (irritability, neuropathy)
- Anemia with basophilic stippling
- Wrist/foot drop

Why other options are incorrect:

- Asbestosis: Lung fibrosis (no effect on heme synthesis)
- Silicosis: Occupational lung disease
- Mercury poisoning: Neurological and renal toxicity, not porphyrin metabolism

QUESTION

Heavy metal chelators are all EXCEPT:

A Penicillamine

B Oxime

CORRECT

C BAL

D Trientine

RIGHT ANSWER

OK B. Oxime

WHY THIS IS RIGHT

Heavy metal chelators are agents that bind toxic metals and form stable complexes that can be excreted from the body, thereby reducing metal toxicity.

Examples include:

- Penicillamine - used in copper poisoning and Wilson disease.
- BAL (Dimercaprol) - used in arsenic, mercury, and gold poisoning.
- Trientine - another copper chelator used in Wilson disease.

Oximes (e.g., pralidoxime) are not heavy-metal chelators. They are used in organophosphate poisoning to reactivate acetylcholinesterase by removing the phosphate group from the inhibited enzyme.

QUESTION

Which of the following connective tissue degradation disorder is associated with aortic aneurysm?

A Ehlers-Danlos Syndrome

B Marfan's Syndrome

CORRECT

C Atherosclerosis

D Scurvy

RIGHT ANSWER

OK B. Marfan's Syndrome

WHY THIS IS RIGHT

Marfan syndrome is a connective tissue disorder caused by mutation in the fibrillin-1 (FBN1) gene, leading to defective elastic fiber formation in connective tissues. Weakness of the aortic media predisposes to aortic root dilatation, aneurysm, and dissection, which is a major cause of mortality in these patients.

It is also characterized by tall stature, long limbs (arachnodactyly), lens dislocation, and skeletal abnormalities.

Why not other options:

- Ehlers-Danlos syndrome: Defect in collagen synthesis causing hyperextensible skin and joint hypermobility; vascular complications may occur in rare types but aortic aneurysm classically associated with Marfan syndrome.
- Atherosclerosis: Causes arterial narrowing and plaque formation, not primarily a connective-tissue degradation disorder.
- Scurvy: Vitamin C deficiency causing defective collagen synthesis leading to bleeding gums and poor wound healing, not aortic aneurysm.

QUESTION

Which of the following deficiencies is associated with sexual maturation defect and hypogonadism?

A Selenium

B Zn

CORRECT

C Cu

D Iron

RIGHT ANSWER

OK B. Zn

WHY THIS IS RIGHT

Zinc deficiency is associated with delayed sexual maturation, hypogonadism, and impaired reproductive function. Zinc plays an important role in DNA synthesis, cell division, and hormone metabolism, including the normal function of the hypothalamic-pituitary-gonadal axis.

Deficiency of zinc can lead to delayed puberty, impaired spermatogenesis, decreased testosterone levels, growth retardation, poor wound healing, and decreased immunity.

Therefore, zinc is essential for normal sexual development and reproductive health, and its deficiency can result in hypogonadism and delayed sexual maturation.

QUESTION

A Patient had dietary Cu deficiency, which is required by lysyl oxidase to cross link the collagen, what is the diagnosis of the patient?

A Menkes Disease

CORRECT

B Scoliosis

C Ehlers Danlos syndrome

D Scurvy

RIGHT ANSWER

A. Menkes Disease

WHY THIS IS RIGHT

Copper is an essential cofactor for the enzyme lysyl oxidase, which catalyzes the oxidative deamination of lysine and hydroxylysine residues in collagen and elastin, allowing cross-linking of connective tissue fibers. Role of lysyl oxidase:

- Requires Cu^{2+} (copper) as a cofactor
- Forms cross-links between collagen and elastin fibers
- Provides tensile strength and structural stability to connective tissue

Menkes disease:

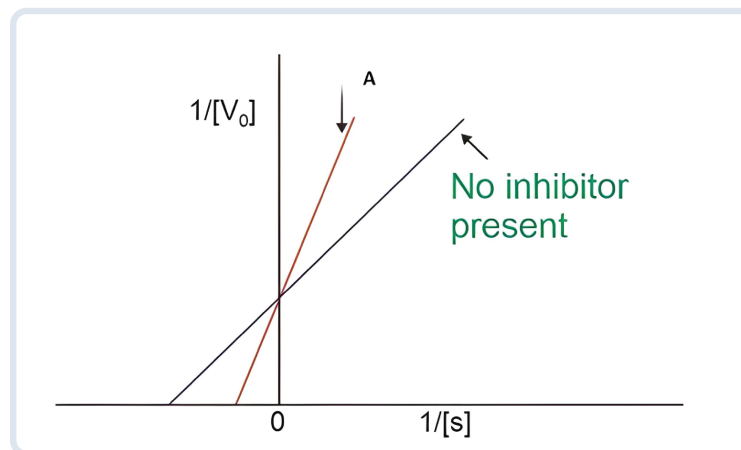
- X-linked recessive disorder
- Caused by mutation in ATP7A gene -> defective copper transport
- Leads to systemic copper deficiency and reduced lysyl oxidase activity
- Weak connective tissue
- Kinky (steely) hair
- Neurodegeneration
- Failure to thrive
- Hypopigmentation

Differentiation from other options:

- Scurvy -> Vitamin C deficiency -> impaired hydroxylation of collagen
- Ehlers-Danlos syndrome -> collagen structure/synthesis defects
- Scoliosis -> spinal curvature disorder, not enzyme-related

QUESTION

Identify the type of inhibition exhibited by A.



A Competitive inhibition

CORRECT

B Uncompetitive inhibition

C Mixed inhibition

D Non-competitive inhibition

RIGHT ANSWER

OK A. Competitive inhibition

WHY THIS IS RIGHT

The graph represents competitive inhibition, where the inhibitor competes with the substrate for the active site of the enzyme.

Key features in Lineweaver-Burk plot:

- Same Y-intercept ($1/V_{max}$ unchanged)
- Slope increases
- X-intercept shifts toward zero ($\uparrow K_m$)

Interpretation:

- V_{max} remains unchanged $\rightarrow$ inhibition can be overcome by increasing substrate concentration
- K_m increases $\rightarrow$ decreased affinity of enzyme for substrate (more substrate needed)

Why other options are incorrect:

- Uncompetitive inhibition: Parallel lines ($\downarrow K_m$ and $\downarrow V_{max}$)
- Non-competitive inhibition: $\downarrow V_{max}$, K_m unchanged (same X-intercept)
- Mixed inhibition: Both K_m and V_{max} change (lines intersect off axes)

QUESTION

Detoxification of Xenobiotics for phase 1 reaction (Hydroxylation) require monooxygenase/cytochrome P-450 along with:

A NADPH

CORRECT

B NADH

C FAD

D FADH₂

RIGHT ANSWER

OK

A. NADPH

WHY THIS IS RIGHT

Phase I detoxification (functionalization reactions) of xenobiotics mainly involves oxidation reactions such as hydroxylation, catalyzed by the cytochrome P450 monooxygenase system present in the smooth endoplasmic reticulum of liver cells.

In this system, NADPH acts as the reducing agent and provides electrons through NADPH-cytochrome P450 reductase, which allows molecular oxygen to be split, with one oxygen atom incorporated into the substrate (hydroxylation) and the other reduced to water.

Why other options are incorrect:

- NADH: Mainly involved in mitochondrial electron transport and not the primary reducing agent for cytochrome P450 reactions.
- FAD / FADH₂: These act as prosthetic groups in some enzymes but are not the direct electron donors for the cytochrome P450 monooxygenase system.

QUESTION

| The active site of enzyme is complimentary to:

A Transition state

B Allosteric activator

C Substrate

CORRECT

D Product

RIGHT ANSWER

OK C. Substrate

WHY THIS IS RIGHT

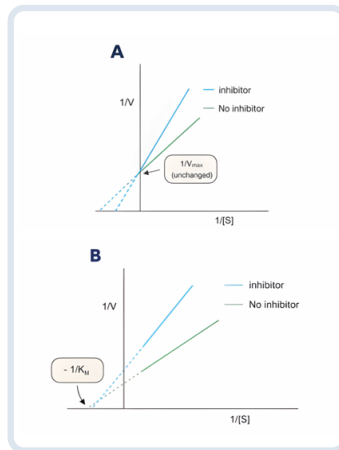
The active site of an enzyme is a specific region where the substrate binds to form the enzyme-substrate complex. The shape and chemical properties of the active site are complementary to the substrate, allowing specific recognition and binding, which enables the enzyme to catalyze the reaction efficiently.

Why other options are incorrect:

- Transition state: The enzyme stabilizes the transition state during catalysis, but the active site is primarily designed to bind the substrate initially.
- Allosteric activator: These molecules bind to allosteric sites, not the active site.
- Product: Products are formed after the reaction and typically have lower affinity for the active site, allowing their release.

QUESTION

Inhibitor A and B are



A Competitive and non-competitive

CORRECT

B Uncompetitive and non-competitive

C Non-competitive and uncompetitive

D Non-competitive and competitive

RIGHT ANSWER

OK

A. Competitive and non-competitive

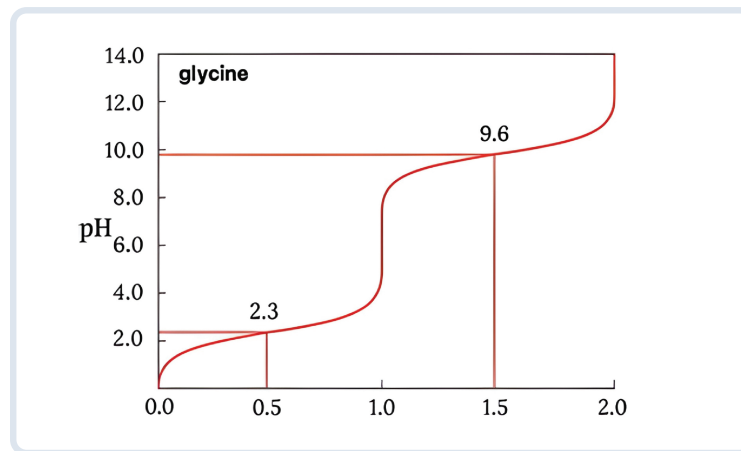
WHY THIS IS RIGHT

Inhibitor A is a competitive inhibitor, and Inhibitor B is a non-competitive inhibitor.

Type of inhibition	Constant Parameter	Variable Parameter	Lineweaver-Burk Plot Intersection
Competitive Inhibition	V_{max} (unchanged)	K_M (increased) V_{max}	On Y-axis (same V_{max})
Non-Competitive Inhibition	K_M (unchanged)	(decreased)	On X-axis (same K_M)

QUESTION

In the given glycine titration curve, calculate or identify the pH at which glycine predominantly exists in its zwitterionic form?



A 4.4

B 6

CORRECT

C 9

D 2.2

RIGHT ANSWER

OK B. 6

WHY THIS IS RIGHT

Glycine's $pI = (pK_a 2.3 + pK_a 9.6) \div 2 \approx 6$, where it exists mainly as a zwitterion with zero net charge.

At isoelectric pH:

- Proteins have no electrophoretic mobility.
- Minimum solubility -> maximal precipitation.
- Least stability, prone to aggregation.
- Do not bind to ion-exchange columns.

QUESTION

A family that consumes a diet predominantly consisting of polished rice develops symptoms of muscle weakness and difficulty in movement. Which of the following nutrients should be administered as prophylaxis?

A Calcium

B Thiamine

CORRECT

C Vitamin D

D Iron

RIGHT ANSWER

OK

B. Thiamine

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$1a” here and does not include a written explanation for this item.

QUESTION

A 45-year-old woman presents with fatigue, pallor, and glossitis. Laboratory evaluation reveals macrocytic anemia. Serum studies show elevated homocysteine levels, while methylmalonic acid (MMA) levels are normal. Which of the following is the most likely underlying cause?

A Vitamin B12 deficiency

B Folate deficiency

CORRECT

C Iron deficiency anemia

D Thalassemia minor

RIGHT ANSWER

OK B. Folate deficiency

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$1b” here and does not include a written explanation for this item.

QUESTION

Which of the following vitamin deficiencies is most likely to cause lactic acidosis?

A Niacin

B Vitamin C

C Thiamine

CORRECT

D Riboflavin

RIGHT ANSWER

OK C. Thiamine

WHY THIS IS RIGHT

Thiamine (Vitamin B1) is an essential cofactor for several enzymes involved in aerobic carbohydrate metabolism, particularly:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase

When thiamine is deficient, the pyruvate dehydrogenase reaction is impaired, preventing the conversion of pyruvate to acetyl-CoA. As a result, pyruvate is instead converted to lactate, leading to accumulation of lactic acid and lactic acidosis.

Why other options are incorrect:

- Niacin: Deficiency causes pellagra (dermatitis, diarrhea, dementia).
- Vitamin C: Deficiency leads to scurvy due to impaired collagen synthesis.
- Riboflavin: Deficiency causes cheilosis, glossitis, and dermatitis, not lactic acidosis.

QUESTION

A woman on anti-depressants presents with bleeding. She gives a history of bulky stools which sticks to the pan. Which of the following vitamin deficiency can cause bleeding in this condition?

A Vitamin A

B Vitamin D

C Vitamin E

D **Vitamin K**

CORRECT

RIGHT ANSWER

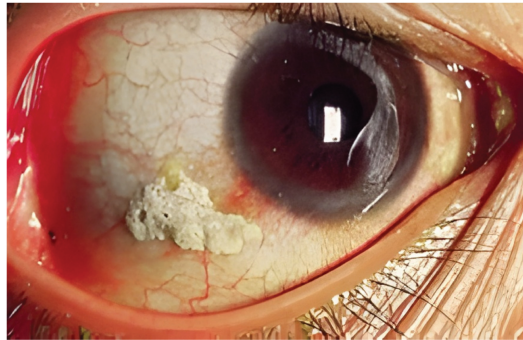
OK **D. Vitamin K**

WHY THIS IS RIGHT

Bulky, foul-smelling stools that stick to the pan indicate fat malabsorption (steatorrhea). Since vitamin K is a fat-soluble vitamin (A, D, E, K), its absorption is impaired in conditions causing fat malabsorption. Vitamin K is essential for γ -carboxylation of clotting factors II, VII, IX, and X, as well as proteins C and S. Deficiency leads to impaired coagulation and presents with bleeding tendencies. Additionally, some antidepressants (e.g., SSRIs) can increase bleeding risk by affecting platelet function, further worsening the bleeding in vitamin K deficiency.

QUESTION

| Condition shown below is due to which vitamin deficiency?



CORRECT

A Vitamin A

B Vitamin B

C Vitamin C

D Vitamin K

RIGHT ANSWER

OK A. Vitamin A

WHY THIS IS RIGHT

Image shows Bitot spots-foamy, whitish patches on the conjunctiva, which are characteristic of vitamin A deficiency and part of the spectrum of xerophthalmia.

- Vitamin A is essential for maintenance of epithelial integrity and mucin production in the conjunctiva.
- Deficiency leads to keratinization of conjunctival epithelium and accumulation of debris -> forming Bitot spots.
- It also impairs rhodopsin formation, causing night blindness.

Spectrum of Vitamin A deficiency:

1. Night blindness
2. Conjunctival xerosis
3. Bitot spots
4. Corneal xerosis
5. Keratomalacia (corneal softening -> blindness)

Why other options are incorrect:

- Vitamin B: Causes glossitis, neuropathy, anemia
- Vitamin C: Causes scurvy (bleeding gums, poor wound healing)
- Vitamin K: Causes bleeding due to clotting factor deficiency

QUESTION

FAD status in body is determined by?

- A** Thiamine
- B** **Glutathione reductase**
- C** Hexokinase
- D** Transketolase

CORRECT

RIGHT ANSWER

- OK** **B. Glutathione reductase**

WHY THIS IS RIGHT

FAD (Flavin adenine dinucleotide) is derived from riboflavin (vitamin B2) and functions as a cofactor in various redox reactions.

The functional status of FAD in the body is assessed by measuring the activity of glutathione reductase in red blood cells. This enzyme requires FAD as a cofactor to convert oxidized glutathione (GSSG) to reduced glutathione (GSH).

- In riboflavin (FAD) deficiency, glutathione reductase activity decreases.
- When FAD is added in vitro, enzyme activity increases (activation coefficient), indicating deficiency.

Why other options are incorrect:

- Thiamine: Assessed by transketolase activity (not FAD).
- Hexokinase: Involved in glycolysis, not used for vitamin assessment.
- Transketolase: Used to assess thiamine (vitamin B1) status.

QUESTION

A known patient of tuberculosis, on INH for the past 3 months complains of numbness, tingling and burning sensation in all the extremities. What could be vitamin deficiency?

A Vitamin B6

CORRECT

B Vitamin B12

C Vitamin B1

D Vitamin B3

RIGHT ANSWER

OK

A. Vitamin B6

WHY THIS IS RIGHT

Isoniazid (INH) therapy in tuberculosis can cause vitamin B6 (pyridoxine) deficiency, leading to peripheral neuropathy (numbness, tingling, burning sensation).

- INH forms complexes with pyridoxine and increases its excretion
- This leads to ↓ availability of pyridoxal phosphate (active B6)
- Pyridoxal phosphate is required for neurotransmitter synthesis (e.g., GABA)

Deficiency results in:

- Peripheral neuropathy
- Paresthesia (glove and stocking pattern)

Prevention: Always give prophylactic vitamin B6 (pyridoxine) with INH therapy

Why other options are incorrect:

- Vitamin B12: Causes neuropathy but not specifically linked to INH
- Vitamin B1 (Thiamine): Causes beriberi/Wernicke's
- Vitamin B3 (Niacin): Causes pellagra (dermatitis, diarrhea, dementia)

QUESTION

Identify the vitamin deficiency:



A Vitamin A

B Vitamin D

CORRECT

C Vitamin B3

D Vitamin B1

RIGHT ANSWER

OK B. Vitamin D

WHY THIS IS RIGHT

The image shows bowed legs (genu varum) in a child, which is a classical feature of rickets due to vitamin D deficiency.

- Vitamin D is essential for calcium and phosphate absorption from the intestine
- Deficiency -> ↓ calcium absorption -> defective mineralization of growing bones
- Leads to soft, weak bones that bend under body weight -> bowed legs

Other features of rickets:

- Rachitic rosary (beading at costochondral junctions)
- Widened wrists
- Frontal bossing
- Delayed growth

Why other options are incorrect:

- Vitamin A: Night blindness, xerophthalmia
- Vitamin B3 (Niacin): Pellagra (dermatitis, diarrhea, dementia)
- Vitamin B1 (Thiamine): Beriberi (neuropathy, cardiac issues)

QUESTION

A child presents with thinning of hair and scaling of the scalp. On taking further history, it is found that she consumes raw eggs in her diet regularly. The given manifestations are due to the deficiency of which of the following vitamins?

- A** Niacin
- B** Pyridoxine
- C** Biotin
- D** Thiamine

CORRECT

RIGHT ANSWER

OK C. Biotin

WHY THIS IS RIGHT

Consumption of raw eggs leads to biotin (vitamin B7) deficiency due to the presence of avidin, a protein in egg white that binds biotin and prevents its absorption.

Biotin functions as a coenzyme for carboxylation reactions:

- Pyruvate carboxylase
- Acetyl-CoA carboxylase
- Propionyl-CoA carboxylase

Clinical features of biotin deficiency:

- Alopecia (hair thinning/loss)
- Dermatitis (scaly skin, especially scalp)
- Neurological symptoms (depression, lethargy)

QUESTION

A patient presenting with bleeding gums and easy bruisability was diagnosed with scurvy. What is the biochemical basis of scurvy?

- A** Low calcium
- B** Defective collagen formation
- C** Inhibition of clotting factors
- D** Increased keratinization of epithelium

CORRECT

RIGHT ANSWER

- OK** **B. Defective collagen formation**

WHY THIS IS RIGHT

Scurvy is caused by vitamin C (ascorbic acid) deficiency, leading to defective collagen synthesis. Vitamin C is required for hydroxylation of proline and lysine residues in collagen via prolyl and lysyl hydroxylase. This hydroxylation is essential for:

- Stabilization of collagen triple helix
- Proper cross-linking of collagen fibers

Clinical features explained:

- Bleeding gums & petechiae -> weak vascular connective tissue
- Poor wound healing
- Bone pain (in children)

Why other options are incorrect:

- Low calcium: Causes bone disorders (rickets/osteomalacia)
- Inhibition of clotting factors: Seen in vitamin K deficiency
- Increased keratinization: Seen in vitamin A deficiency

QUESTION

A child was brought with pedal edema and cheilosis. Further investigations revealed cardiomegaly. What is the vitamin deficiency associated with this clinical presentation?

A Riboflavin

B Thiamine

CORRECT

C Pyridoxine

D Niacin

RIGHT ANSWER

OK

B. Thiamine

WHY THIS IS RIGHT

Thiamine (Vitamin B1) deficiency leads to beriberi, which can present with both cardiac and peripheral manifestations. Types relevant here:

- Wet beriberi:

- o Cardiomegaly, high-output cardiac failure
- o Edema (pedal edema)

- Dry beriberi:

- o Peripheral neuropathy

Thiamine is a cofactor for key enzymes in energy metabolism:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase

Cheilosis: Though classically seen in riboflavin deficiency, it can coexist in malnutrition, but the cardiomegaly + edema strongly points to thiamine deficiency.

Why other options are incorrect:

- Riboflavin: Causes cheilosis and glossitis but not cardiomegaly
- Pyridoxine: Causes neuropathy and sideroblastic anemia
- Niacin: Causes pellagra (dermatitis, diarrhea, dementia)

QUESTION

A chronic alcoholic patient presented to the casualty with confusion, ataxia, and painful eye movements. What could be the vitamin deficiency seen in this patient?

A Vitamin B deficiency

CORRECT

B Vitamin A deficiency

C Vitamin D deficiency

D Vitamin C deficiency

RIGHT ANSWER

A. Vitamin B deficiency

WHY THIS IS RIGHT

A chronic alcoholic presenting with confusion, ataxia, and painful/abnormal eye movements (ophthalmoplegia) is exhibiting the classic triad of Wernicke encephalopathy, caused by thiamine (Vitamin B1) deficiency.

Thiamine is a cofactor for key enzymes in glucose metabolism:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase

Deficiency $\rightarrow$ $\downarrow$ ATP production $\rightarrow$ neuronal energy failure, especially in brain regions like mammillary bodies and periaqueductal gray

Clinical triad:

- Confusion
- Ataxia
- Ophthalmoplegia (eye movement abnormalities)

Why other options are incorrect:

- Vitamin A: Vision problems (night blindness)
- Vitamin D: Bone disorders
- Vitamin C: Scurvy (bleeding gums)

QUESTION

A family consumes only polished rice. Which of the following combination of vitamin deficiency and enzymatic defect will be present in this family?

A Riboflavin - Glutathione Reductase

B Thiamine - Transketolase

CORRECT

C Thiamine - Transaminase

D Riboflavin - Transketolase

RIGHT ANSWER

B. Thiamine - Transketolase

WHY THIS IS RIGHT

A diet consisting predominantly of polished rice leads to thiamine (Vitamin B1) deficiency, because the outer bran layer (rich in thiamine) is removed during polishing.

Thiamine (as thiamine pyrophosphate, TPP) is a cofactor for several enzymes, including:

- Transketolase (in the HMP shunt)
- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase

Among these, transketolase activity in RBCs is commonly used to assess thiamine status.

Why other options are incorrect:

- Riboflavin - Glutathione reductase: Correct pair, but riboflavin deficiency is not the primary issue with polished rice diet
- Thiamine - Transaminase: Transaminases require vitamin B6 (pyridoxine)
- Riboflavin - Transketolase: Transketolase is dependent on thiamine, not riboflavin

QUESTION

A patient presented with diarrhea, dermatitis, and dementia. Which of the following vitamin deficiencies would you suspect in this patient?

A Vitamin B3 deficiency

CORRECT

B Vitamin B1 deficiency

C Vitamin B6 deficiency

D Vitamin B12 deficiency

RIGHT ANSWER

OK **A. Vitamin B3 deficiency**

WHY THIS IS RIGHT

The triad of diarrhea, dermatitis, and dementia is characteristic of pellagra, caused by niacin (Vitamin B3) deficiency.

- Precursor of NAD⁺ and NADP⁺
- Essential for redox reactions and energy metabolism
- Affects tissues with high turnover -> skin, GI tract, and brain

Clinical features (3 D's):

- Dermatitis: Photosensitive, hyperpigmented rash (Casal necklace)
- Diarrhea: Due to GI mucosal atrophy
- Dementia: Neurological impairment

(+ 4th D: Death if untreated)

Why other options are incorrect:

- Vitamin B1: Beriberi, Wernicke's encephalopathy
- Vitamin B6: Neuropathy, sideroblastic anemia
- Vitamin B12: Megaloblastic anemia + neurological deficits (no dermatitis/diarrhea triad)

QUESTION

| Which of the following zymogens requires gamma-glutamyl carboxylase?

A Factors II, VIII, IX, X

B Factors II, VII, IX, and X

CORRECT

C Factors II, VII, IX, XI

D Factors II, VIII, X, XI

RIGHT ANSWER

OK **B. Factors II, VII, IX, and X**

WHY THIS IS RIGHT

γ -Glutamyl carboxylase is a vitamin K-dependent enzyme that catalyzes the γ -carboxylation of glutamate residues on certain clotting factors. This modification allows these factors to bind calcium (Ca^{2+}) and attach to phospholipid surfaces, which is essential for normal coagulation cascade function. Vitamin K-dependent clotting factors:

- Factor II (Prothrombin)
- Factor VII
- Factor IX
- Factor X

(+ Proteins C and S)

Clinical relevance:

- Vitamin K deficiency or warfarin therapy $\rightarrow$ impaired γ -carboxylation $\rightarrow$ bleeding tendency
- First factor to be affected: Factor VII (shortest half-life $\rightarrow$ $\uparrow$ PT earliest)

Why other options are incorrect:

- Factor VIII: Not vitamin K dependent
- Factor XI: Not vitamin K dependent

QUESTION

A 12-year-old adolescent presents with progressive joint pain, easy fatigability, and weakness. On examination, the patient has marfanoid habitus and mild developmental delay. Laboratory evaluation shows elevated homocysteine levels with decreased cysteine levels. Which vitamin supplementation is most appropriate in this condition?

A Vitamin B6 (Pyridoxine)

CORRECT

B Vitamin B2 (Riboflavin)

C Vitamin B1 (Thiamine)

D Vitamin B7 (Biotin)

RIGHT ANSWER

OK **A. Vitamin B6 (Pyridoxine)**

WHY THIS IS RIGHT

In homocystinuria, elevated homocysteine with low cysteine indicates a defect in the transsulfuration pathway, specifically the enzyme cystathionine β -synthase (CBS).

- Vitamin B6 (PLP) is a cofactor for CBS
- Required for conversion of:

Homocysteine $\rightarrow$ Cystathionine $\rightarrow$ Cysteine

- CBS deficiency $\rightarrow$ $\uparrow$ homocysteine and $\downarrow$ cysteine
- Leads to vascular damage, thrombosis, skeletal abnormalities

Giving Vitamin B6 enhances residual enzyme activity, reducing homocysteine levels

Clinical features of homocystinuria:

- Marfanoid habitus
- Lens subluxation (downward)
- Thromboembolism
- Intellectual disability

Why other options are incorrect:

- Vitamin B2: FAD-related enzymes
- Vitamin B1: Carbohydrate metabolism
- Vitamin B7 (Biotin): Carboxylation reactions

QUESTION

| Which vitamin can be deficient in a patient who has undergone gastrectomy?

A Vitamin A

B Vitamin B12

CORRECT

C Vitamin C

D Vitamin D

RIGHT ANSWER

OK B. Vitamin B12

WHY THIS IS RIGHT

Vitamin B12 (cobalamin) deficiency is common after gastrectomy due to loss of intrinsic factor (IF) production.

- Parietal cells of stomach produce intrinsic factor
- IF is essential for absorption of vitamin B12 in the terminal ileum
- Gastrectomy -> ↓ intrinsic factor -> impaired B12 absorption

Consequences:

- Megaloblastic anemia
- Neurological manifestations (subacute combined degeneration)
 - o Paresthesia
 - o Loss of vibration & position sense
 - o UMN signs

Why other options are incorrect:

- Vitamin A: Fat-soluble, absorbed in intestine
- Vitamin C: Absorbed directly, not IF-dependent
- Vitamin D: Fat-soluble, not affected by intrinsic factor

QUESTION

A 45-year-old woman with Rheumatoid arthritis is started on low-dose methotrexate therapy. After a few weeks, she develops oral ulcers and mild bone marrow suppression. Which of the following is most directly reduced due to this mechanism?

A Thymidine monophosphate (TMP) CORRECT

B Guanosine monophosphate (GMP)

C Cytidine monophosphate (CMP)

D Adenosine monophosphate (AMP)

RIGHT ANSWER

OK **A. Thymidine monophosphate (TMP)**

WHY THIS IS RIGHT

Methotrexate is a folate antagonist that inhibits the enzyme dihydrofolate reductase (DHFR).

- Blocks conversion of dihydrofolate (DHF) → tetrahydrofolate (THF)
- THF is required for one-carbon transfer reactions
- Impaired synthesis of thymidylate (dTMP) from dUMP
- Leads to ↓ DNA synthesis, especially in rapidly dividing cells

Clinical relevance:

- Used in cancer chemotherapy, autoimmune diseases (RA)
- Causes megaloblastic anemia as a side effect

Why other options are incorrect:

- GMP, AMP: Purine synthesis also affected indirectly but main tested effect is dTMP inhibition
- CMP: Pyrimidine synthesis not directly dependent on folate in this step

QUESTION

A patient presented with follicular hyperkeratosis on the extensor aspect of the forearm. What is the diagnosis?

A Darier's disease

B Phrynoderma

CORRECT

C Eruptive xanthoma

D Folliculitis

RIGHT ANSWER

B. Phrynoderma

WHY THIS IS RIGHT

Phrynoderma ("toad skin") is characterized by follicular hyperkeratosis on extensor surfaces (arms, thighs), most commonly due to vitamin A deficiency.

- Vitamin A is essential for normal epithelial differentiation
- Deficiency -> abnormal keratinization of hair follicles -> rough, dry papules

Clinical features:

- Rough, keratotic papules over extensor surfaces
- Dry, coarse skin
- May be associated with night blindness

Why other options are incorrect:

- Darier's disease: Genetic disorder with greasy papules (seborrheic areas)
- Eruptive xanthoma: Yellow papules due to hypertriglyceridemia
- Folliculitis: Infection of hair follicles (pus-filled lesions)

QUESTION

In the shown image, which one of the following is a source of vitamin B1?



A A

CORRECT

B B

C C

D D

RIGHT ANSWER

OK A. A

WHY THIS IS RIGHT

Vitamin B1 (Thiamine) is primarily obtained from whole grains, cereals, and pulses (unpolished rice, legumes)-represented by option A in the image.

Thiamine (as thiamine pyrophosphate, TPP) acts as a cofactor for:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase (HMP shunt)

Deficiency leads to:

- Beriberi (dry: neuropathy, wet: cardiac failure)
- Wernicke-Korsakoff syndrome (in alcoholics)

QUESTION

The treatment given to the patient presenting with a history of binge alcohol intake and symptoms of convulsions, altered sensorium, and plasma glucose level of 45 mg/dL is:

A 25% dextrose

B Fomepizole

C Thiamine

D Thiamine followed by dextrose

CORRECT

RIGHT ANSWER

OK

D. Thiamine followed by dextrose

WHY THIS IS RIGHT

In a chronic alcoholic with hypoglycemia (glucose 45 mg/dL) and altered sensorium, the correct management is thiamine administration before glucose.

- Chronic alcoholics are commonly thiamine (Vitamin B1) deficient
- Thiamine is required for glucose metabolism (pyruvate dehydrogenase, etc.)
- Giving glucose first increases metabolic demand and can precipitate or worsen Wernicke encephalopathy

Correct sequence: Thiamine first -> then dextrose

- Thiamine deficiency -> impaired cerebral energy metabolism
- Glucose without thiamine -> accumulation of lactate -> neuronal injury

Why other options are incorrect:

- 25% dextrose alone: Can worsen neurological damage
- Fomepizole: Used in methanol/ethylene glycol poisoning
- Thiamine alone: Does not correct hypoglycemia

QUESTION

Which micronutrient deficiency causes anemia, bleeding gums, and white line of Frenkel on X-ray?

A Copper

B Vitamin C

CORRECT

C Iron

D Zinc

RIGHT ANSWER

B. Vitamin C

WHY THIS IS RIGHT

The triad of anemia, bleeding gums, and “white line of Frankel” on X-ray is characteristic of vitamin C (ascorbic acid) deficiency -> scurvy.

Vitamin C is essential for hydroxylation of proline and lysine in collagen via:

- Prolyl hydroxylase
- Lysyl hydroxylase

Deficiency -> defective collagen synthesis -> weak connective tissue.

Clinical features explained:

- Bleeding gums & petechiae: Fragile capillaries due to weak collagen
- Anemia: Due to chronic bleeding + ↓ iron absorption (vitamin C enhances iron absorption)
- White line of Frankel (dense metaphyseal line): Due to defective osteoid formation in growing bones

Radiological signs in scurvy:

- White line of Frankel
- Pelkan spurs
- Scorbutic rosary

Why other options are incorrect:

- Copper: Causes anemia and neurological issues, not bleeding gums + Frankel line
- Iron: Causes anemia only
- Zinc: Causes dermatitis and poor wound healing

QUESTION

A patient present with symptoms of dermatitis, diarrhea, and cognitive decline. Which micronutrient deficiency is most likely responsible?



A Niacin

CORRECT

B Thiamine

C Pyridoxine

D Retinol

RIGHT ANSWER

OK A. Niacin

WHY THIS IS RIGHT

The image shows Casal necklace—a photosensitive hyperpigmented dermatitis around the neck, which is characteristic of pellagra due to niacin (Vitamin B3) deficiency.

Niacin is a precursor of NAD⁺ and NADP⁺, essential for oxidation-reduction reactions and energy metabolism.

Deficiency → impaired cellular energy production, especially in skin, GI tract, and brain.

Classic triad (3 D's):

- Dermatitis (photosensitive, Casal necklace)
- Diarrhea
- Dementia

(+ Death if untreated)

Why other options are incorrect:

- Thiamine: Beriberi, Wernicke's encephalopathy
- Pyridoxine: Neuropathy, sideroblastic anemia
- Retinol (Vitamin A): Night blindness, xerophthalmia

QUESTION

Which amino acid will not be synthesized due to B12 deficiency in a patient with history of ileectomy with elevated homocysteine levels, increased methylmalonyl CoA and peripheral neuropathy?

A Methionine

CORRECT

B Cysteine

C Glutathione

D Tryptophan

RIGHT ANSWER

OK
A. Methionine

WHY THIS IS RIGHT

Vitamin B12 deficiency (e.g., after ileectomy, where B12 is absorbed) leads to impairment of two key reactions:

1. Homocysteine → Methionine (via methionine synthase, B12-dependent)
2. Methylmalonyl-CoA → Succinyl-CoA (via methylmalonyl-CoA mutase)

Biochemical consequences:

- ↑ Homocysteine (cannot convert to methionine)
- ↓ Methionine synthesis
- ↑ Methylmalonyl-CoA → contributes to neurological damage

Clinical relevance:

- Peripheral neuropathy (due to myelin damage)
- Megaloblastic anemia
- Seen in ileal resection (ileectomy) due to impaired B12 absorption

Why other options are incorrect:

- Cysteine: Formed from homocysteine (via B6-dependent pathway), not directly dependent on B12
- Glutathione: Synthesized from glutamate, cysteine, glycine
- Tryptophan: Essential amino acid (not synthesized in body)

QUESTION

A non-alcoholic hypertensive patient on hydrochlorothiazide therapy presents with cerebellar ataxia, peripheral neuropathy, symmetrical tingling sensations, cardiac failure, and pedal edema. Deficiency of which of the following vitamins or nutrients can cause similar clinical features?

A Selenium

B Thiamine

CORRECT

C Vit B12

D Vit B2

RIGHT ANSWER

B. Thiamine

WHY THIS IS RIGHT

The constellation of cerebellar ataxia, peripheral neuropathy, tingling, cardiac failure, and pedal edema is characteristic of thiamine (Vitamin B1) deficiency, which presents as a combination of dry beriberi (neuropathy) and wet beriberi (cardiac failure + edema).

Chronic diuretic use can increase urinary loss of thiamine, predisposing to deficiency

Thiamine (as TPP) is a cofactor for:

- Pyruvate dehydrogenase
- α -ketoglutarate dehydrogenase
- Transketolase

Deficiency -> impaired ATP production, especially affecting:

- Nervous system -> neuropathy, ataxia
- Heart -> cardiomyopathy, heart failure, edema

Why other options are incorrect:

- Selenium: Cardiomyopathy (Keshan disease) but no neuropathy triad
- Vitamin B12: Neuropathy without cardiac failure/edema
- Vitamin B2: Cheilosis, glossitis, dermatitis

QUESTION

A patient presents with fatigue, gum bleeding, and multiple perifollicular hemorrhages on the lower limbs, along with a bleeding tendency. These findings are most consistent with a deficiency of which enzyme activity?

A Prolyl hydroxylase

CORRECT

B Lysyl oxidase

C Tyrosinase

D Alkaline phosphatase

RIGHT ANSWER

OK

A. Prolyl hydroxylase

WHY THIS IS RIGHT

The clinical features-bleeding gums, perifollicular hemorrhages, easy bruising, and fatigue-are classic for scurvy (vitamin C deficiency).

Vitamin C is required as a cofactor for:

- Prolyl hydroxylase
- Lysyl hydroxylase

These enzymes catalyze hydroxylation of proline and lysine residues in collagen, which is essential for:

- Stability of the collagen triple helix
- Proper cross-linking of collagen fibers

Why other options are incorrect:

- Lysyl oxidase: Involved in collagen cross-linking but is copper-dependent, not vitamin C dependent
- Tyrosinase: Involved in melanin synthesis
- Alkaline phosphatase: Associated with bone metabolism

QUESTION

| Which of the following statements regarding folate deficiency is FALSE?

- A** High homocysteine and normal methyl malonic acid
- B** If given at the diagnosis of pregnancy - can prevent neural tube defects CORRECT
- C** Folate supplementation is required in children with chronic hemolytic anemia
- D** Vitamin B12 deficiency should be corrected before supplementing folate

RIGHT ANSWER

- OK** **B. If given at the diagnosis of pregnancy - can prevent neural tube defects**

WHY THIS IS RIGHT

Folate is essential for DNA synthesis and neural tube development. To prevent neural tube defects (NTDs) such as spina bifida and anencephaly, folic acid supplementation must begin before conception and during early pregnancy (first 4 weeks), when neural tube closure occurs. Therefore, starting folate only at the time of pregnancy diagnosis may be too late to prevent NTDs, making that statement false.

Other statements are true:

- Folate deficiency: ↑ homocysteine with normal methylmalonic acid.
- Chronic hemolytic anemia: Increased RBC turnover -> higher folate requirement.
- Vitamin B12 deficiency should be corrected before folate therapy to avoid masking B12 deficiency and worsening neurological damage.

QUESTION

Korsakoff psychosis is true except?

1. Amnesia
2. Confabulation
3. Ophthalmoplegia
4. Ataxia

A 1, 2, 3

B 2, 3, 4

C 1, 3

D 3, 4

CORRECT

RIGHT ANSWER

OK D. 3, 4

WHY THIS IS RIGHT

Korsakoff psychosis is a chronic neuropsychiatric disorder caused by thiamine (vitamin B₁) deficiency, most commonly seen in chronic alcoholism. It is characterized by anterograde and retrograde amnesia with confabulation (fabrication of stories to fill memory gaps). Ophthalmoplegia and ataxia are features of Wernicke encephalopathy, the acute stage of thiamine deficiency.

QUESTION

In folate deficiency, the False statement is:

- A** High homocysteine and normal methylmalonic acid
- B** Hemolytic anemia is a feature CORRECT
- C** B12 supplementation is recommended along with Folate
- D** Purine and pyrimidine synthesis are affected

RIGHT ANSWER

- OK** **B. Hemolytic anemia is a feature**

WHY THIS IS RIGHT

Folate deficiency causes impaired DNA synthesis due to defective purine and thymidylate (pyrimidine) synthesis, leading to megaloblastic anemia. It is characterized by elevated homocysteine levels with normal methylmalonic acid (unlike vitamin B₁₂ deficiency).

Hemolytic anemia is not a typical feature; instead, folate deficiency produces megaloblastic anemia due to ineffective erythropoiesis. Vitamin B₁₂ is often supplemented with folate to avoid masking an underlying B₁₂ deficiency and preventing neurological complications.

QUESTION

A 52-year-old strict vegetarian presents with progressive numbness in feet, gait instability, and memory changes. Examination reveals loss of vibration and position sense with spasticity in lower limbs. Blood tests show macrocytic anemia. He is started on folic acid therapy, after which his anemia improves, but neurological symptoms worsen.

What is the most likely mechanism for this deterioration?

A Folate trap leading to methyl-THF accumulation

CORRECT

B Depletion of S-adenosylmethionine (SAM)

C Pernicious anemia

D Homocysteine accumulation

RIGHT ANSWER

OK **A. Folate trap leading to methyl-THF accumulation**

WHY THIS IS RIGHT

In vitamin B₁₂ deficiency, folate becomes trapped as methyl-tetrahydrofolate (methyl-THF) due to impaired conversion of homocysteine to methionine, a reaction that requires vitamin B₁₂.

- Methionine synthase requires vitamin B₁₂
- Without B₁₂ -> methyl-THF cannot convert to THF
- Leads to "folate trap" with accumulation of methyl-THF

Clinical Significance:

- Folate supplementation corrects anemia by providing folate for DNA synthesis
- But it does not correct B₁₂ deficiency
- Neurologic damage (due to impaired myelin synthesis) may worsen or continue

QUESTION

Vitamin leading to diversion of 3-hydroxykynurenine toward xanthurenic acid:

CORRECT

A Pyridoxine

B Pantothenic acid

C Thiamine

D Biotin

RIGHT ANSWER

OK

A. Pyridoxine

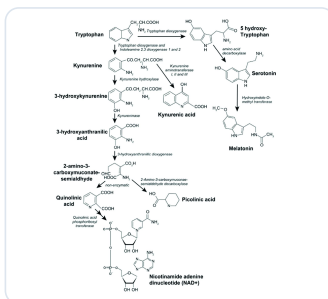
WHY THIS IS RIGHT

Pyridoxine (Vitamin B6) is required as a cofactor in the tryptophan-kynurenine pathway. It is necessary for the enzyme kynureninase, which converts 3-hydroxykynurenine → 3-hydroxyanthranilic acid.

When pyridoxine is deficient:

- The normal pathway is blocked.
- 3-hydroxykynurenine is diverted to form xanthurenic acid.
- Increased urinary excretion of xanthurenic acid occurs.

Can occur with isoniazid therapy, which interferes with pyridoxine metabolism.



QUESTION

| Which type of PCR is suitable for identifying multiple causative agents?

A Conventional PCR

B Real-time PCR

C **Multiplex PCR**

CORRECT

D Nested PCR

RIGHT ANSWER

OK **C. Multiplex PCR**

WHY THIS IS RIGHT

Multiplex PCR is used for simultaneous detection of multiple pathogens in a single reaction by using multiple sets of primers targeting different DNA sequences.

- Principle:

- o Multiple primer pairs -> amplify different targets in one tube
- o Allows identification of multiple causative agents at once

- Advantages:

- o Saves time and cost
- o Requires smaller sample volume
- o Useful in syndromic diagnosis (e.g., respiratory, GI infections)

- Why not others:

- o Conventional PCR: detects single target per reaction
- o Real-time PCR: quantitative, but usually single target (unless multiplexed)
- o Nested PCR: increases sensitivity/specificity, not for multiple agents

QUESTION

A patient presents with multiple colonic polyps and is diagnosed with colorectal carcinoma. There is a strong family history suggestive of Hereditary Non-Polyposis Colorectal Cancer (HNPCC). Which DNA repair mechanism is most likely defective in this condition?

A Nucleotide excision repair

B Mismatch repair

CORRECT

C Base excision repair

D Double-strand break repair

RIGHT ANSWER

B. Mismatch repair

WHY THIS IS RIGHT

Hereditary Non-Polyposis Colorectal Cancer (HNPCC), also called Lynch syndrome, is caused by defects in the DNA mismatch repair (MMR) system. This repair mechanism normally corrects base-base mismatches and small insertion/deletion errors that occur during DNA replication.

Common genes involved:

- MLH1
- MSH2
- MSH6
- PMS2

Why other options are incorrect:

- Nucleotide excision repair: Repairs bulky DNA lesions such as thymine dimers caused by UV radiation (defective in xeroderma pigmentosum).
- Base excision repair: Corrects small base modifications such as deamination or oxidation.
- Double-strand break repair: Repairs DNA breaks through homologous recombination or non-homologous end joining.

QUESTION

| When a codon is changed to stop codon, then it is:

A Stop codon mutation

B **Non-sense mutation**

CORRECT

C Mis sense Mutation

D Silent Mutation

RIGHT ANSWER

OK **B. Non-sense mutation**

WHY THIS IS RIGHT

A nonsense mutation is a type of point mutation in which a codon for an amino acid is converted into a stop codon (UAA, UAG, or UGA). This results in premature termination of protein synthesis, producing a truncated and usually nonfunctional protein.

Why other options are incorrect:

- Stop codon mutation: Not a standard classification term used in genetics.
- Missense mutation: One amino acid is replaced by another (no stop codon formed).
- Silent mutation: Codon change does not alter the amino acid due to redundancy of the genetic code.

QUESTION

| True about telomeres is:

- A** Increased telomere length with ageing
- B** Gradually decreasing length of telomeres with ageing CORRECT
- C** Telomeres have coding sequences
- D** Telomerase is found in somatic cells

RIGHT ANSWER

- OK** **B. Gradually decreasing length of telomeres with ageing**

WHY THIS IS RIGHT

Telomeres are repetitive non-coding DNA sequences (TTAGGG repeats) present at the ends of chromosomes that protect them from degradation and fusion.

With each cell division, due to the end-replication problem, telomeres gradually shorten, leading to:

- Cellular aging (senescence)
- Limited replicative capacity of somatic cells

Telomerase is an enzyme that adds telomeric repeats to chromosome ends, thereby maintaining telomere length. However:

- It is active mainly in germ cells, stem cells, and cancer cells
- It is absent or very low in most somatic cells

Why other options are incorrect:

- Increased telomere length with ageing: Opposite is true.
- Telomeres have coding sequences: They are non-coding repetitive DNA.
- Telomerase is found in somatic cells: Generally absent or minimal in somatic cells.

QUESTION

| DNA packing is done by?

A Histone

CORRECT

B Glycoprotein

C Nucleic acid

D Helicases

RIGHT ANSWER

OK

A. Histone

WHY THIS IS RIGHT

DNA packaging in eukaryotic cells is carried out by histone proteins, which help condense long DNA molecules into a compact, organized structure within the nucleus.

- DNA wraps around histone octamers (2 each of H2A, H2B, H3, H4) forming nucleosomes
- Nucleosomes further coil to form chromatin fibers and ultimately chromosomes

Functions of histones:

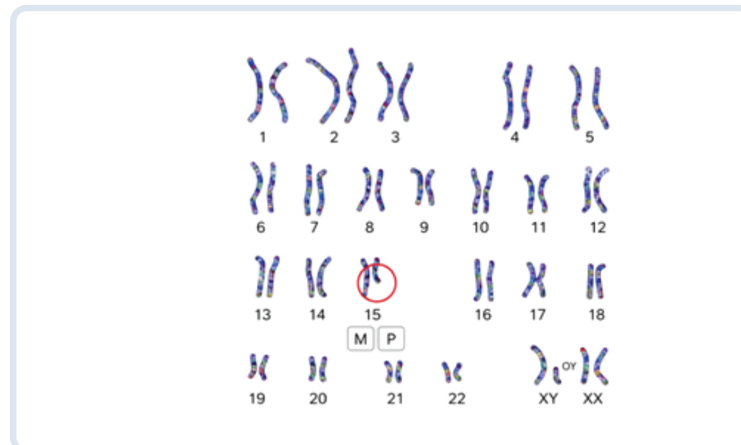
- DNA compaction (fits long DNA into nucleus)
- Regulation of gene expression (via histone modification like acetylation, methylation)

Why other options are incorrect:

- Glycoprotein: Not involved in DNA packaging
- Nucleic acid: DNA itself, not responsible for packing
- Helicases: Enzymes that unwind DNA during replication, not packaging

QUESTION

What is the diagnosis of the marked karyotype?



A Prader-Willi syndrome

CORRECT

B DiGeorge syndrome

C Cri du Chat syndrome

D Angelman syndrome

RIGHT ANSWER

OK **A. Prader-Willi syndrome**

WHY THIS IS RIGHT

The karyotype shows a defect involving chromosome 15, consistent with Prader-Willi syndrome, caused by deletion of the paternal allele of chromosome 15 (15q11-q13) due to genomic imprinting.

- Paternal gene deletion or maternal uniparental disomy
- Imprinting -> only one parental allele is active
- Loss of paternal gene expression -> disease

Clinical features:

- Hyperphagia -> obesity
- Hypotonia in infancy
- Intellectual disability
- Hypogonadism

Why other options are incorrect:

- DiGeorge syndrome: 22q11 deletion
- Cri du Chat: 5p deletion (cat-like cry)
- Angelman syndrome: Maternal chromosome 15 deletion -> ataxia, seizures, "happy puppet"

QUESTION

A 7-year-old child presents with severe photosensitivity, freckling over sun-exposed areas, and recurrent skin lesions since early childhood. There is a history of progressive neurological decline. On examination, multiple hyperpigmented macules and areas of skin atrophy are noted on the face and arms. The underlying defect involves impaired repair of UV-induced thymine dimers. Which DNA repair mechanism is primarily affected in this condition?

- A** Base excision repair
- B** Double-strand DNA break repair
- C** Mismatch repair

D Nucleotide excision repair

CORRECT

RIGHT ANSWER

OK D. Nucleotide excision repair

WHY THIS IS RIGHT

UV radiation causes DNA damage in the form of pyrimidine (thymine) dimers, which distort the DNA helix. These lesions are repaired by the nucleotide excision repair (NER) pathway.

1. Recognition of DNA distortion (thymine dimers)
2. Excision of the damaged DNA segment
3. DNA polymerase fills the gap
4. DNA ligase seals the strand

Clinical relevance:

- Defect in NER -> Xeroderma pigmentosum (XP)
- Presents with photosensitivity, exaggerated skin rash on sun exposure, and early skin cancers

Why other options are incorrect:

- Base excision repair: Repairs small base changes (oxidation, deamination)
- Double-strand break repair: Repairs DNA breaks (HR, NHEJ)
- Mismatch repair: Corrects replication errors (base mismatches)

QUESTION

A mother brought her 14-year-old mentally retarded child with complaints of joint pain and an irresistible urge to bite his fingers and lips. On investigation, his serum uric acid level was found to be elevated. The child is likely suffering from which disorder?

A Xanthine oxidase deficiency

B Thymidylate synthetase

C Lesch-Nyhan syndrome

CORRECT

D CPSII defect

RIGHT ANSWER

OK C. Lesch-Nyhan syndrome

WHY THIS IS RIGHT

The clinical features-intellectual disability, self-mutilation (biting lips/fingers), joint pain, and hyperuricemia-are characteristic of Lesch-Nyhan syndrome.

- Deficiency of HGPRT (Hypoxanthine-guanine phosphoribosyltransferase)
- A key enzyme in the purine salvage pathway
- Impaired salvage of hypoxanthine and guanine -> increased degradation to uric acid
- ↑ PRPP and ↓ IMP/GMP -> ↑ de novo purine synthesis -> further uric acid production

Clinical features explained:

- Hyperuricemia -> gout, joint pain
- Neurological dysfunction -> intellectual disability
- Self-mutilation behavior (classic hallmark)

Why other options are incorrect:

- Xanthine oxidase deficiency: ↓ uric acid (xanthinuria), not hyperuricemia
- Thymidylate synthetase defect: Affects DNA synthesis
- CPS II defect: Affects pyrimidine synthesis

QUESTION

A frameshift mutation occurs due to the insertion of a new nucleotide at the 4th position of an mRNA sequence containing 900 nucleotides. What is the most likely outcome of this mutation?

A No change in the final protein

B Partial protein production

C Complete change in protein production

CORRECT

D No change due to RNA editing

RIGHT ANSWER

OK **C. Complete change in protein production**

WHY THIS IS RIGHT

A frameshift mutation occurs when nucleotides are inserted or deleted in numbers not divisible by 3, altering the reading frame of the mRNA.

In this case, insertion of a nucleotide at the 4th position (early in the sequence) causes:

- Shift in the reading frame from that point onward
- All subsequent codons are altered
- Often leads to a premature stop codon
- Produces a completely abnormal (nonfunctional) protein

Why other options are incorrect:

- No change in protein: Seen in silent mutations, not frameshift
- Partial protein production: More typical of nonsense mutation (premature stop without frame shift)
- No change due to RNA editing: RNA editing does not correct frameshift mutations

QUESTION

A child presents with multiple freckles, severe photosensitivity, and early-onset skin cancers. What is the primary underlying molecular defect responsible for this condition?

A Nucleotide excision repair defect

CORRECT

B Mismatch repair defect

C Base excision repair defect

D Homologous recombination defect

RIGHT ANSWER

OK

A. Nucleotide excision repair defect

WHY THIS IS RIGHT

The clinical features-multiple freckles, severe photosensitivity, and early-onset skin cancers-are characteristic of Xeroderma Pigmentosum (XP).

Underlying molecular defect:

- Defective nucleotide excision repair (NER) pathway
- This pathway normally repairs UV-induced DNA damage, especially pyrimidine (thymine) dimers

Pathophysiology:

- UV radiation -> formation of thymine dimers
- NER defect -> inability to remove damaged DNA
- Accumulation of mutations -> high risk of skin cancers at an early age

Why other options are incorrect:

- Mismatch repair defect: Seen in Lynch syndrome (HNPCC)
- Base excision repair defect: Repairs small base damage (oxidation, deamination)
- Homologous recombination defect: Seen in BRCA mutations (breast/ovarian cancer)

QUESTION

Which PCR technique is considered the most appropriate method for syndromic diagnosis of pathogens causing meningitis?

- A Nested PCR
- B Multiplex PCR
- C RT-PCR
- D Uniplex PCR

CORRECT

RIGHT ANSWER

OK

B. Multiplex PCR

WHY THIS IS RIGHT

No explanation was provided in the source quiz.

PCR Type	Location	Function
Multiplex PCR		Uses multiple primer sets in one reaction to detect several pathogens simultaneously.
Nested PCR		Two-step PCR using inner + outer primers to give high sensitivity/specificity for a single low-copy organism.
RT-PCR (qPCR)		Real-time fluorescent PCR giving quantitative detection of one target per reaction (unless multiplexed).
Uniplex PCR		Standard PCR that detects only one organism per reaction, needing separate tubes for each target.

QUESTION

| Which technique is used to identify auto antibodies?

A Direct immunofluorescence

B ELISA

C Indirect immunofluorescence

CORRECT

D FISH

RIGHT ANSWER

OK C. Indirect immunofluorescence

WHY THIS IS RIGHT

Indirect immunofluorescence (IIF) is commonly used to detect circulating autoantibodies in patient serum. In this technique, the patient's serum is incubated with a substrate containing the target antigen (e.g., cells or tissue). If autoantibodies are present, they bind to the antigen. A fluorescent-labeled secondary antibody (anti-human IgG) is then added to detect these bound antibodies, producing fluorescence under a microscope.

Why other options are incorrect:

- Direct immunofluorescence: Detects antigen-antibody complexes deposited in tissues, not circulating autoantibodies in serum.
- ELISA: Can detect antibodies, but the classical and widely used screening method for many autoantibodies (e.g., ANA patterns) is indirect immunofluorescence.
- FISH (Fluorescence in situ hybridization): Used to detect DNA or chromosomal abnormalities, not antibodies.

QUESTION

In which of the genetic material, Number of Adenine will not be equal to that of Thymine:

- A E. coli
- B Mitochondrial DNA of human
- C HIV
- D Herpes virus

CORRECT

RIGHT ANSWER

OK C. HIV

WHY THIS IS RIGHT

According to Chargaff's rule, in double-stranded DNA the amount of adenine (A) equals thymine (T) because they pair together (A-T). This rule applies to organisms with double-stranded DNA genomes, such as E. coli, human mitochondrial DNA, and herpes virus.

HIV, however, contains a single-stranded RNA genome, not double-stranded DNA. Since base pairing is not fixed in single-stranded nucleic acids, adenine does not necessarily equal thymine (or uracil). Therefore, Chargaff's rule does not apply, making HIV the correct answer.

QUESTION

Which Biochemical method is exploited for monoclonal antibody production by hybridoma technique?

- A** Glycolytic pathway
- B** Pentose Phosphate pathway
- C** Kreb's cycle
- D** Purine salvage pathway

CORRECT

RIGHT ANSWER

- OK** **D. Purine salvage pathway**

WHY THIS IS RIGHT

The hybridoma technique for producing monoclonal antibodies uses HAT medium (Hypoxanthine-Aminopterin-Thymidine) for selection. In this medium, cells must use the purine salvage pathway to synthesize nucleotides because aminopterin blocks the de novo purine synthesis pathway.

Only hybridoma cells (fusion of antibody-producing B cells with myeloma cells) survive, as they possess the necessary enzymes (e.g., HGPRT) to utilize the purine salvage pathway, allowing selective growth and production of monoclonal antibodies.

Why not other options:

- Glycolytic pathway: Involved in glucose breakdown for ATP, not nucleotide synthesis or HAT selection.
- Pentose phosphate pathway: Produces NADPH and ribose-5-phosphate, but does not bypass the aminopterin block in HAT medium.
- Krebs cycle: Part of energy metabolism in mitochondria, unrelated to nucleotide salvage used in hybridoma selection.

QUESTION

FISH is used to detect all of the following except?

A Translocation

B Amplification

C Deletion

D Point mutation

CORRECT

RIGHT ANSWER

OK **D. Point mutation**

WHY THIS IS RIGHT

Fluorescence in situ hybridization (FISH) is a cytogenetic technique that uses fluorescent DNA probes to detect large chromosomal abnormalities such as translocations, deletions, duplications, and gene amplifications. However, FISH cannot detect point mutations because these involve single nucleotide changes, which are too small to be identified by probe-based chromosomal hybridization. Detection of point mutations requires molecular techniques such as PCR or DNA sequencing.

QUESTION

| Which of the following is not associated with post-transcriptional modification?

- A** Methylation
- B** Endonuclease cleavage
- C** Capping
- D** Glycosylation

CORRECT

RIGHT ANSWER

- OK** **D. Glycosylation**

WHY THIS IS RIGHT

Post-transcriptional modifications occur after RNA is synthesized from DNA and are essential for converting pre-mRNA into mature mRNA. The major modifications include:

- 5' Capping: Addition of a 7-methylguanosine cap at the 5' end, which protects mRNA from degradation and helps in ribosome binding.
- Endonuclease cleavage and splicing: Removal of introns and joining of exons to form mature mRNA.
- Methylation: Certain bases in RNA may be methylated during RNA processing.

Glycosylation is not a post-transcriptional modification. It is a post-translational modification of proteins that occurs in the endoplasmic reticulum and Golgi apparatus, where carbohydrate groups are added to proteins.

QUESTION

Which type of mutation results in the reversal to the wild type of phenotype when the mutant gene is suppressed?

A Frameshift mutation of coding gene

B Addition of another normal gene

C Deletion of the mutant gene

D Mutation of tRNA

CORRECT

RIGHT ANSWER

OK

D. Mutation of tRNA

WHY THIS IS RIGHT

A suppressor mutation can restore the wild-type phenotype of a mutant gene without correcting the original mutation. One common mechanism involves a mutation in tRNA that allows it to recognize altered codons (such as stop codons or frameshift codons) and insert an amino acid during translation.

These mutant tRNAs (suppressor tRNAs) compensate for the defective coding sequence, permitting translation to continue and partially or completely restoring protein function.

Thus, mutation of tRNA can suppress the effect of the original mutation and revert the phenotype toward wild type, even though the original DNA mutation remains present.

QUESTION

Which of the following is the best investigation for aneuploidy?

- A. In Situ Hybridization
- B. Conventional cytogenetics
- C. PCR
- D. Sanger sequencing

A A only

B A & B

CORRECT

C C & D

D A, B, C and D

RIGHT ANSWER

OK B. A & B

WHY THIS IS RIGHT

Aneuploidy refers to an abnormal number of chromosomes (e.g., trisomy or monosomy). The most appropriate investigations are karyotyping (conventional cytogenetics) and fluorescence in situ hybridization (FISH) because they directly evaluate chromosome number and structure.

- Conventional cytogenetics (karyotyping) visualizes the entire chromosome set and is the gold standard for detecting numerical abnormalities such as trisomy 21, trisomy 18, and monosomy X.
- In situ hybridization (FISH) uses fluorescent probes targeting specific chromosomes and provides rapid detection of aneuploidy, commonly used in prenatal and interphase cells.

PCR and Sanger sequencing are primarily used to detect gene-level mutations or small sequence variations, not whole-chromosome number abnormalities. Therefore, they are not ideal for detecting aneuploidy.

QUESTION

| All are used in recombinant DNA technology (genetic engineering) except?

- A** Phosphatases
- B** CRISPR Cas9
- C** **Isomerases**
- D** Terminal transferase

CORRECT

RIGHT ANSWER

OK **C. Isomerases**

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$19” here and does not include a written explanation for this item.

QUESTION

A scientist is culturing a virus using the RT-PCR technique and performs 26 amplification cycles. After how many cycles would the DNA quantity be approximately half of what is present at the 26th cycle?

A 27

B 25

CORRECT

C 13

D 10

RIGHT ANSWER

OK B. 25

WHY THIS IS RIGHT

In PCR amplification, the amount of DNA doubles with each cycle (exponential amplification). the DNA quantity at 25 cycles is approximately half of that at 26 cycles.

$$\text{DNA amount} = 2^n$$

- At 26 cycles $\rightarrow 2^{26}$ copies
- At 25 cycles $\rightarrow 2^{25}$ copies

Since

$$2^{25} = \frac{1}{2} \times 2^{26}$$

QUESTION

Match the following:

Column 1	Column 2
1. Southern blotting	a. Proteins
2. Northern blotting	b. Enzymes
3. Western blotting	c. RNA
4. Eastern blotting	d. DNA

- A

1-a, 2-d, 3-c, 4-b
- B

1-d, 2-c, 3-a, 4-b

CORRECT
- C

1-a, 2-b, 3-c, 4-d
- D

1-b, 2-c, 3-d, 4-a

RIGHT ANSWER

OK

B. 1-d, 2-c, 3-a, 4-b

WHY THIS IS RIGHT

No explanation was provided in the source quiz.

Technique	Detects	Key Use
Southern blot	DNA	Gene mapping, detecting mutations, RFLP analysis, prenatal diagnosis
Northern blot	RNA	Measures gene expression (mRNA levels), differentiates active vs. silent genes
Western blot	Proteins	Confirms HIV , Lyme disease, detects specific antibodies/proteins
Eastern blot	Enzymes/post-translational modifications (e.g., glycosylation)	Used to study protein modifications important in signaling pathways

QUESTION

| Which of the following is not an example of aneuploidy?

A Klinefelter syndrome

B Patau syndrome

C Down syndrome

D Bloom syndrome

CORRECT

RIGHT ANSWER

OK **D. Bloom syndrome**

WHY THIS IS RIGHT

Aneuploidy refers to an abnormal number of chromosomes due to nondisjunction during meiosis, resulting in gain or loss of individual chromosomes.

Examples of Aneuploidy

- Down syndrome: Trisomy 21
- Patau syndrome: Trisomy 13
- Klinefelter syndrome: 47, XXY (sex chromosome aneuploidy)

Bloom syndrome is not an aneuploidy; it is a chromosomal instability disorder caused by mutation in the BLM gene (DNA helicase) leading to defective DNA repair and increased sister chromatid exchanges.

QUESTION

A 58-year-old patient with a known malignancy undergoes molecular analysis of tumor cells. RNA sequencing reveals transcripts that are longer than normal, with inclusion of additional exon sequences not seen in healthy tissue. Which of the following mechanisms best explains this finding?

- A** Polyadenylation
- B** Alternative RNA splicing
- C** Gene amplification
- D** Upstream promoter modification

CORRECT

RIGHT ANSWER

- OK** **B. Alternative RNA splicing**

WHY THIS IS RIGHT

Alternative RNA splicing allows a single gene to produce multiple mRNA transcripts with different exon combinations, leading to variation in transcript length and protein isoforms.

Role in Cancer:

- Cancer cells often exhibit abnormal alternative splicing.
- This can generate longer or altered mRNA transcripts.
- These variants may promote tumor growth, survival, and metastasis.

Why not other options:

- A) Polyadenylation: Adds a poly-A tail at the 3' end but does not significantly change transcript length of coding regions.
- C) Gene amplification: Increases the number of gene copies, not the length of each RNA transcript.
- D) Upstream promoter modification: Affects transcription initiation and expression level, not mRNA length.

QUESTION

A researcher treats cultured human cells with a histone acetyltransferase (HAT) inhibitor and observes rapid epigenetic changes. Within minutes, there is reduced transcriptional activity and chromatin appears more compact. Which of the following molecular changes is most likely to be seen immediately?

A Increased H3K9me2/3

CORRECT

B Increased H3K9 acetylation

C Chromatin relaxation

D DNA methylation

RIGHT ANSWER

OK

A. Increased H3K9me2/3

WHY THIS IS RIGHT

- HAT inhibition → ↓ histone acetylation → chromatin condensation
- Associated with repressive marks (↑ H3K9 methylation)
- Acetylation = active chromatin; methylation (H3K9me2/3) = silenced chromatin

Histone acetylation relaxes chromatin and promotes gene transcription, whereas histone deacetylation leads to chromatin condensation and gene repression.

Effect of Histone Acetylase (HAT) Inhibition:

- ↓ Histone acetylation
- Chromatin becomes more condensed
- Leads to repressive histone marks, such as H3K9 methylation (H3K9me2/3)

QUESTION

| Which of the following correctly depicts the sequence of steps in the PCR?

A Denaturation -> Annealing -> Extension

CORRECT

B Denaturation -> Annealing -> Extension -> Hybridization

C Annealing -> Extension -> Denaturation

D Extension -> Denaturation -> Annealing

RIGHT ANSWER

OK **A. Denaturation -> Annealing -> Extension**

WHY THIS IS RIGHT

Polymerase Chain Reaction (PCR) is a laboratory technique used to amplify a specific segment of DNA, producing millions of copies from a small DNA sample.

Sequence of steps in PCR

1. Denaturation ($\approx 94-98^{\circ}\text{C}$)

- Double-stranded DNA separates into two single strands due to breaking of hydrogen bonds.

2. Annealing ($\approx 50-65^{\circ}\text{C}$)

- Primers bind to complementary sequences on the single-stranded DNA template.

3. Extension / Elongation ($\approx 72^{\circ}\text{C}$)

- DNA polymerase synthesizes new DNA strands by adding nucleotides complementary to the template.

These steps are repeated for multiple cycles, resulting in exponential amplification of the target DNA.